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- (71) Applicant (for all designated States except US): CURA-GEN CORPORATION [US/US]; 11th floor, 555 Long Wharf Drive, Branford, CT 06511 (US).
- (72) Inventors; and
- (75) Inventors/Applicants (for US only): SHIMKETS, Richard, A. [US/US]; 191 Lete Street, West Haven, CT

06516 (US). LEACH, Martin [GB/US]; 884 School Street, Webster, MA 01570 (US).

- (74) Agent: ELRIFI, Ivor, R.; Mintz, Levin, Cohn, Ferris, Glovsky and Popeo PC, One Financial Center, Boston, MA 02111 (US).
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NUCLEIC ACIDS CONTAINING SINGLE NUCLEOTIDE POLYMORPHISMS AND METHODS OF USE THEREOF

FIELD OF THE INVENTION

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The invention relates generally to nucleic acids and polypeptides and in particular to the identification of human single nucleotide polymorphisms based on at least one gene product that was not previously described.

BACKGROUND OF THE INVENTION

Sequence polymorphism-based analysis of nucleic acid is generally based on alterations in nucleic acid sequences between related individuals. This analysis has been widely used in a variety of genetic, diagnostic, and forensic applications. For example, polymorphism analyses are used in identity and paternity analysis, and in genetic mapping studies.

Several different types of polymorphisms in nucleic acid have been described. One such type of variation is a restriction fragment length polymorphism (RFLP). RFLPS can create or delete a recognition sequence for a restriction endonuclease in one nucleic acid relative to a second nucleic acid. The result of the variation is in an alteration the relative length of restriction enzyme generated DNA fragments in the two nucleic acids.

Other polymorphisms take the form of short tandem repeats (STR) sequences, which are also referred to as variable numbers of tandem repeat (VNTR) sequences. STR sequences typically that include tandem repeats of 2, 3, or 4 nucleotide sequences that are present in a nucleic acid from one individual but absent from a second, related individual at the corresponding genomic location.

Other polymorphisms take the form of single nucleotide variations, termed single nucleotide polymorphisms (SNPs), between individuals. A SNP can, in some instances, be referred to as a "cSNP" to denote that the nucleotide sequence containing the SNP originates as a cDNA.

SNPs can arise in several ways. A single nucleotide polymorphism may arise due to a substitution of one nucleotide for another at the polymorphic site. Substitutions can be transitions or transversions. A transition is the replacement of one purine nucleotide by

another purine nucleotide, or one pyrimidine by another pyrimidine. A transversion is the replacement of a purine by a pyrimidine, or the converse.

Single nucleotide polymorphisms can also arise from a deletion of a nucleotide or an insertion of a nucleotide relative to a reference allele. Thus, the polymorphic site is a site at which one allele bears a gap with respect to a single nucleotide in another allele. Some SNPs occur within, or near genes. One such class includes SNPs falling within regions of genes encoding for a polypeptide product. These SNPs may result in an alteration of the amino acid sequence of the polypeptide product and give rise to the expression of a defective or other variant protein. Such variant products can, in some cases result in a pathological condition, e.g., genetic disease. Examples of genes in which a polymorphism within a coding sequence gives rise to genetic disease include sickle cell anemia and cystic fibrosis. Other SNPs do not result in alteration of the polypeptide product. Of course, SNPs can also occur in noncoding regions of genes.

SNPs tend to occur with great frequency and are spaced uniformly throughout the genome. The frequency and uniformity of SNPs means that there is a greater probability that such a polymorphism will be found in close proximity to a genetic locus of interest.

SUMMARY OF THE INVENTION

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The invention is based in part on the discovery of single nucleotide polymorphisms (SNPs) in regions of human DNA.

Accordingly, in one aspect, the invention provides nucleic acid sequences comprising nucleic acid segments of both publicly known and novel genes, including the polymorphic site. The segments can be DNA or RNA, and can be single- or double-stranded. Preferred segments include a biallelic polymorphic site.

The invention further provides allele-specific oligonucleotides that hybridize to a segment of a fragment shown in Table 1, column 4, or its complement. These oligonucleotides can be probes or primers. Also provided are isolated nucleic acids comprising a sequence shown in Table 1, column 4, in which the polymorphic site within the sequence is occupied by a base other than the reference bases shown in Table 1, columns 5 and 6.

The invention further provides a method of analyzing a nucleic acid from an individual. The method determines which base is present at any one of the polymorphic sites shown in Table 1. Optionally, a set of bases occupying a set of polymorphic sites shown in Table 1 is determined. This type of analysis can be performed on a number of individuals, who are tested for the presence of a disease phenotype.

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In another aspect, the invention provides an isolated polynucleotide which includes one or more of the SNPs described herein. The polynucleotide can be, e.g., a nucleotide sequence which includes one or more of the polymorphic sequences shown in Table 1 and which includes a polymorphic sequence, or a fragment of the polymorphic sequence, as long as it includes the polymorphic site. The polynucleotide may alternatively contain a nucleotide sequence which includes a sequence complementary to one or more of these sequences, or a fragment of the complementary nucleotide sequence, provided that the fragment includes a polymorphic site in the polymorphic sequence.

The polynucleotide can be, e.g., DNA or RNA, and can be between about 10 and about 100 nucleotides, e.g., 10-90, 10-75, 10-51, 10-40, or 10-30, nucleotides in length.

In preferred embodiments, the polymorphic site in the polymorphic sequence includes a nucleotide other than the nucleotide listed in Table 1, column 5 for the polymorphic sequence, e.g., the polymorphic site includes the nucleotide listed in Table 1, column 6 for the polymorphic sequence.

In other embodiments, the complement of the polymorphic site includes a nucleotide other than the complement of the nucleotide listed in Table 1, column 5 for the complement of the polymorphic sequence, e.g., the complement of the nucleotide listed in Table 1, column 6 for the polymorphic sequence.

In some embodiments, the polymorphic sequence is associated with a polypeptide related to one of the protein families disclosed herein. For example, the nucleic acid may be associated with a polypeptide related to angiopoietin, 4-hydroxybutyrate dehydrogenase, or any of the other proteins identified in Table 1, column 10.

In another aspect, the invention provides an isolated allele-specific oligonucleotide that hybridizes to a first polynucleotide containing a polymorphic site. The first polynucleotide can be, e.g., a nucleotide sequence comprising one or more polymorphic

sequences recited in Table 1, provided that the polymorphic sequence includes a nucleotide other than the nucleotide recited in Table 1, column 5 for the polymorphic sequence. Alternatively, the first polynucleotide can be a nucleotide sequence that is a fragment of the polymorphic sequence, provided that the fragment includes a polymorphic site in the polymorphic sequence, or a complementary nucleotide sequence which includes a sequence complementary to one or more polymorphic sequences in Table 1, provided that the complementary nucleotide sequence includes a nucleotide other than the complement of the nucleotide recited in Table 1, column 5. The first polynucleotide may in addition include a nucleotide sequence that is a fragment of the complementary sequence, provided that the fragment includes a polymorphic site in the polymorphic sequence.

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In some embodiments, the oligonucleotide does not hybridize under stringent conditions to a second polynucleotide. The second polynucleotide can be, e.g., (a) a nucleotide sequence comprising one or more polymorphic sequences in Table 1, wherein the polymorphic sequence includes the nucleotide listed in Table 1, column 5 for the polymorphic sequence; (b) a nucleotide sequence that is a fragment of any of the polymorphic sequences; (c) a complementary nucleotide sequence including a sequence complementary to one or more polymorphic sequences disclosed herein in Table 1; and (d) a nucleotide sequence that is a fragment of the complementary sequence, provided that the fragment includes a polymorphic site in the polymorphic sequence.

The oligonucleotide can be, e.g., between about 10 and about 100 bases in length. In some embodiments, the oligonucleotide is between about 10 and 75 bases, 10 and 51 bases, 10 and about 40 bases, or about 15 and 30 bases in length.

The invention also provides a method of detecting a polymorphic site in a nucleic acid. The method includes contacting the nucleic acid with an oligonucleotide that hybridizes to a polymorphic sequence selected shown in Table 1, or its complement, provided that the polymorphic sequence includes a nucleotide other than the nucleotide recited in Table 1, column 5 for the polymorphic sequence, or the complement includes a nucleotide other than the complement of the nucleotide recited in Table 1, column 5. The method also includes determining whether the nucleic acid and the oligonucleotide hybridize. Hybridization of the oligonucleotide to the nucleic acid sequence indicates the presence of the polymorphic site in the nucleic acid.

In preferred embodiments, the oligonucleotide does not hybridize to the polymorphic sequence when the polymorphic sequence includes the nucleotide recited in Table 1, column 5 for the polymorphic sequence, or when the complement of the polymorphic sequence includes the complement of the nucleotide recited in Table 1, column 5 for the polymorphic sequence.

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The oligonucleotide can be, e.g., between about 10 and about 100 bases in length. In some embodiments, the oligonucleotide is between about 10 and 75 bases, 10 and 51 bases, 10 and about 40 bases, or about 15 and 30 bases in length.

In some embodiments, the polymorphic sequence identified by the oligonucleotide is associated with a nucleic acid encoding polypeptide related to one of the protein families disclosed herein, the polymorphic sequence is associated with a polypeptide related to one of the protein families disclosed herein. For example, the nucleic acid may be associated with a polypeptide related to angiopoietin, 4-hydroxybutyrate dehydrogenase, or any of the other proteins identified in Table 1, column 10.

In a further aspect, the invention provides a method of determining the relatedness of a first and second nucleic acid. The method includes providing a first nucleic acid and a second nucleic acid and contacting the first nucleic acid and the second nucleic acid with an oligonucleotide that hybridizes to a polymorphic sequence selected disclosed in Table 1, or its complement, provided that the polymorphic sequence includes a nucleotide other than the nucleotide recited in Table 1, column 5 for the polymorphic sequence, or the complement includes a nucleotide other than the complement of the nucleotide recited in Table 1, column 5. The method also includes determining whether the first nucleic acid and the second nucleic acid hybridize to the oligonucleotide, and comparing hybridization of the first and second nucleic acids to the oligonucleotide. Hybridization of first and second nucleic acids to the nucleic acid indicates the first and second subjects are related.

In preferred embodiments, the oligonucleotide does not hybridize to the polymorphic sequence when the polymorphic sequence includes the nucleotide recited in Table 1, column 5 for the polymorphic sequence, or when the complement of the polymorphic sequence includes the complement of the nucleotide recited in Table 1, column 5 column for the polymorphic sequence.

The oligonucleotide can be, e.g., between about 10 and about 100 bases in length. In some embodiments, the oligonucleotide is between about 10 and 75 bases, 10 and 51 bases, 10 and about 40 bases, or about 15 and 30 bases in length.

The method can be used in a variety of applications. For example, the first nucleic acid may be isolated from physical evidence gathered at a crime scene, and the second nucleic acid may be obtained is a person suspected of having committed the crime. Matching the two nucleic acids using the method can establishing whether the physical evidence originated from the person.

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In another example, the first sample may be from a human male suspected of being the father of a child and the second sample may be from a child. Establishing a match using the described method can establishing whether the male is the father of the child.

In another aspect, the method includes determining if a sequence polymorphism is the present in a subject, such as a human. The method includes providing a nucleic acid from the subject and contacting the nucleic acid with an oligonucleotide that hybridizes to a polymorphic sequence disclosed in Table 1, or its complement, provided that the polymorphic sequence includes a nucleotide other than the nucleotide recited in Table 1, column 5 for the polymorphic sequence, or the complement includes a nucleotide other than the complement of the nucleotide recited in Table 1, column 5. Hybridization between the nucleic acid and the oligonucleotide is then determined. Hybridization of the oligonucleotide to the nucleic acid sequence indicates the presence of the polymorphism in said subject.

In another aspect, the invention provides an isolated polypeptide comprising a polymorphic site at one or more amino acid residues, and wherein the protein is encoded by a polynucleotide including one of the polymorphic sequences in Table 1, or their complement, provided that the polymorphic sequence includes a nucleotide other than the nucleotide recited in Table 1, column 5 for the polymorphic sequence, or the complement includes a nucleotide other than the complement of the nucleotide recited in Table 1, column 5.

The polypeptide can be, e.g., related to one of the protein families disclosed herein. For example, polypeptide can be related to angiopoietin, 4-hydroxybutyrate dehydrogenase, ATP-dependent RNA helicase, MHC Class I histocompatibility antigen, or phosphoglycerate kinase.

In some embodiments, the polypeptide is translated in the same open reading frame as is a wild type protein whose amino acid sequence is identical to the amino acid sequence of the polymorphic protein except at the site of the polymorphism.

In some embodiments, the polypeptide encoded by the polymorphic sequence, or its complement, includes the nucleotide listed in Table 1, column 6 for the polymorphic sequence, or the complement includes the complement of the nucleotide listed in Table 1, column 6.

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The invention also provides an antibody that binds specifically to a polypeptide encoded by a polynucleotide comprising a nucleotide sequence encoded by a polynucleotide including one or more of the polymorphic sequences in Table 1, or its complement. The polymorphic sequence includes a nucleotide other than the nucleotide recited in Table 1, column 5 for the polymorphic sequence, or the complement includes a nucleotide other than the complement of the nucleotide recited in Table 1, column 5.

In some embodiments, the antibody binds specifically to a polypeptide encoded by a polymorphic sequence which includes the nucleotide listed in Table 1, column 6 for the polymorphic sequence.

Preferably, the antibody does not bind specifically to a polypeptide encoded by a polymorphic sequence which includes the nucleotide listed in Table 1, column 5 for the polymorphic sequence.

The invention further provides a method of detecting the presence of a polypeptide having one or more amino acid residue polymorphisms in a subject. The method includes providing a protein sample from the subject and contacting the sample with the above-described antibody under conditions that allow for the formation of antibody-antigen complexes. The antibody-antigen complexes are then detected. The presence of the complexes indicates the presence of the polypeptide.

The invention also provides a method of treating a subject suffering from, at risk for, or suspected of, suffering from a pathology ascribed to the presence of a sequence polymorphism in a subject, e.g., a human, non-human primate, cat, dog, rat, mouse, cow, pig, goat, or rabbit. The method includes providing a subject suffering from a pathology associated with aberrant expression of a first nucleic acid comprising a polymorphic sequence

shown in Table 1, or its complement, and treating the subject by administering to the subject an effective dose of a therapeutic agent. Aberrant expression can include qualitative alterations in expression of a gene, e.g., expression of a gene encoding a polypeptide having an altered amino acid sequence with respect to its wild-type counterpart. Qualitatively different polypeptides can include, shorter, longer, or altered polypeptides relative to the amino acid sequence of the wild-type polypeptide. Aberrant expression can also include quantitative alterations in expression of a gene. Examples of quantitative alterations in gene expression include lower or higher levels of expression of the gene relative to its wild-type counterpart, or alterations in the temporal or tissue-specific expression pattern of a gene. Finally, aberrant expression may also include a combination of qualitative and quantitative alterations in gene expression.

The therapeutic agent can include, e.g., second nucleic acid comprising the polymorphic sequence, provided that the second nucleic acid comprises the nucleotide present in the wild type allele. In some embodiments, the second nucleic acid sequence comprises a polymorphic sequence which includes nucleotide listed in Table 1, column 5 for the polymorphic sequence.

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Alternatively, the therapeutic agent can be a polypeptide encoded by a polynucleotide comprising polymorphic sequence shown in Table 1, or by a polynucleotide comprising a nucleotide sequence that is complementary to any one of the polymorphic sequences, provided that the polymorphic sequence includes the nucleotide listed in Table 1, column 6 for the polymorphic sequence.

The therapeutic agent may further include an antibody as herein described, or an oligonucleotide comprising a polymorphic sequence shown in Table 1, or by a polymucleotide comprising a nucleotide sequence that is complementary to any one the polymorphic sequences, provided that the polymorphic sequence includes the nucleotide listed in Table 1, column 6 for the polymorphic sequence,

In another aspect, the invention provides an oligonucleotide array comprising one or more oligonucleotides hybridizing to a first polynucleotide at a polymorphic site encompassed therein. The first polynucleotide can be, e.g., a nucleotide sequence comprising one or more polymorphic sequences shown in Table 1; a nucleotide sequence that is a fragment of any of the nucleotide sequence, provided that the fragment includes a

polymorphic site in the polymorphic sequence; a complementary nucleotide sequence comprising a sequence complementary to one or more of the polymorphic sequences; or a nucleotide sequence that is a fragment of the complementary sequence, provided that the fragment includes a polymorphic site in the polymorphic sequence.

In preferred embodiments, the array comprises 10; 100; 1,000; 10,000; 100,000 or more oligonucleotides.

The invention also provides a kit comprising one or more of the herein-described nucleic acids. The kit can include, e.g., polynucleotide which includes one or more of the SNPs described herein. The polynucleotide can be, e.g., a nucleotide sequence which includes one or more of the polymorphic sequences shown in Table 1, and which includes a polymorphic sequence, or a fragment of the polymorphic sequence, as long as it includes the polymorphic site. The polynucleotide may alternatively contain a nucleotide sequence which includes a sequence complementary to one or more of the sequences, or a fragment of the complementary nucleotide sequence, provided that the fragment includes a polymorphic site in the polymorphic sequence.

Alternatively, or in addition, the kit can include the invention provides an isolated allele-specific oligonucleotide that hybridizes to a first polynucleotide containing a polymorphic site. The first polynucleotide can be, e.g., a nucleotide sequence comprising one or more polymorphic sequences shown in Table 1, provided that the polymorphic sequence includes a nucleotide other than the nucleotide recited in Table 1, column 5 for the polymorphic sequence. Alternatively, the first polynucleotide can be a nucleotide sequence that is a fragment of the polymorphic sequence, provided that the fragment includes a polymorphic site in the polymorphic sequence, or a complementary nucleotide sequence which includes a sequence complementary to one or more polymorphic sequences shown in Table 1, provided that the complementary nucleotide sequence includes a nucleotide other than the complement of the nucleotide recited in Table 1, column 6. The first polynucleotide may in addition include a nucleotide sequence that is a fragment of the complementary sequence, provided that the fragment includes a polymorphic site in the polymorphic sequence.

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BRIEF DESCRIPTION OF THE DRAWING

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FIG. 1 illustrates an example of the way in which SNP sites were identified in the present invention.

Unless otherwise defined, all technical and scientific terms used herein have the same meaning as commonly understood by one of ordinary skill in the art to which this invention belongs. Although methods and materials similar or equivalent to those described herein can be used in the practice or testing of the present invention, suitable methods and materials are described below. All publications, patent applications, patents, and other references mentioned herein are incorporated by reference in their entirety. In the case of conflict, the present specification, including definitions, will control. In addition, the materials, methods, and examples are illustrative only and not intended to be limiting.

Other features and advantages of the invention will be apparent from the following detailed description and claims.

DETAILED DESCRIPTION OF THE INVENTION

The invention provides human SNPs in sequences which are transcribed, *i.e.*, are cSNPs. Many SNPs have been identified in genes related to polypeptides of known function. If desired, SNPs associated with various polypeptides can be used together. For example, SNPs can be grouped according to whether they are derived from a nucleic acid encoding a polypeptide related to particular protein family or involved in a particular function. Similarly, SNPs can be grouped according to the functions played by their gene products. Such functions include, structural proteins, proteins from which associated with metabolic pathways fatty acid metabolism, glycolysis, intermediary metabolism, calcium metabolism, proteases, and amino acid metabolism, etc. Specifically, the present invention provides a large number of human cSNP's based on at least one gene product that has not been previously identified. In contrast, and as defined specifically in the following paragraph, the cSNP's involve nucleic acid sequences that are assembled from at least one known sequence.

The present invention describes 651 distinct polymorphic sites, which are summarized in Table 1. Raw traces underlying sequence data were drawn from public databases and from the proprietary database of the Assignee of the present invention. The sequences were obtained by calling the bases from these traces, and included assigning "Phred" quality scores

for each called base. For each allelic set, at the polynucleotide level, four or more nucleotide sequences were identified having at least partial overlap with one another.

As illustrated in FIG. 1, these four or more sequences could be clustered and assembled to make a consensus contig that included an ORF. In this way, the inventors found that the assembled contigs defined associated sets of two, or possibly more than two, alleles defined by a SNP at a particular polymorphic site. In order to be confirmed as a SNP site, the nucleotide change from the consensus sequence had to occur in at least two individual sequences, and had to have a "Phred" score of 23 or higher at the site of the presumed SNP. Furthermore, in a window of 5 bases on either side of the SNP, no more than 50% mismatching with the consensus sequence was allowed. In the assembly leading to each of the contigs defining the allelic set, the SNP alleles occur in polynucleotides found in public databases.

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It was found that the assembled contigs defined associated sets of two, or possibly more than two, alleles defined by an SNP at a particular polymorphic site. These associations were not previously known.

At the level of translation of an ORF contained in the contigs, allelic sets were identified in which one allele defines a known polypeptide sequence that includes the polymorphic site and another polypeptide allele is not previously known. Then, various associations of alleles are possible. For example, it is possible that an allelic pair is defined in a noncoding region of the contig containing an ORF. In such cases the inventors believe that the invention resides in the recognition of the allelic pair; this association has not heretofore been made.

Alternatively, sets of allelic contigs may exist in which the polymorphic site is within an ORF, but does not result in an amino acid change among the allelic polypeptides. Thus, in another embodiment, the polymorphic site resides within an ORF and results in an amino acid change, or a frameshift, among the alleles of the allelic set. In the sets of gene products that fall within this group, at least one of the alleles at the polypeptide level is a known protein. At least one of the remaining allele or alleles in the set, carrying a variant amino acid at the polymorphic site, is a novel polypeptide not heretofore known. The invention resides at least in the recognition of the polymorphic allele as being a variant of the known reference polypeptide.

Table 1 provides information concerning the allelic sequences. One of the sequences may be termed a reference polymorphic sequence, and the corresponding second sequence includes the variant SNP at the polymorphic site. Since the reference polypeptide sequence is already known, the Sequence Listing accompanying this application provides only the sequence of the polymorphic allele, while its SEQ ID NO is provided in the Table. A reference to the SEQ ID NO that corresponds to the translated amino acid sequence is also given. The Table includes thirteen columns that provide descriptive information for each cSNP, each of which occupies one row in the Table. The column headings, and a description of each, are given below.

SNPs disclosed in Table 1 were detected by aligning large numbers of sequences from genetically diverse sources of publicly available mRNA libraries (Clontech). Software designed specifically to look for multiple examples of variant bases differing from a consensus sequence was created and deployed. A criteria of a minimum of 2 occurrences of a sequence differing from the consensus in high quality sequence reads was used to identify an SNP.

The SNPs described herein may be useful in diagnostic kits, for DNA arrays on chips and for other uses that involve hybridization of the SNP.

Specific SNPs may have utility where a disease has already been associated with that gene. Examples of possible disease correlations between the claimed SNPs with members of the genes of each classification are listed below:

Amylases

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Amylase is responsible for endohydrolysis of 1,4-alpha-glucosidic linkages in oligosaccharides and polysaccharides. Variations in amylase gene may be indicative of delayed maturation and of various amylase producing neoplasms and carcinomas.

25 Amyloid

The serum amyloid A (SAA) proteins comprise a family of vertebrate proteins that associate predominantly with high density lipoproteins (HDL). The synthesis of certain members of the family is greatly increased in inflammation. Prolonged elevation of plasma SAA levels, as in chronic inflammation, 15 results in a pathological condition, called amyloidosis, which affects the liver, kidney and spleen and which is characterized by the

highly insoluble accumulation of SAA in these tissues. Amyloid selectively inhibits insulinstimulated glucose utilization and glycogen deposition in muscle, while not affecting adipocyte glucose metabolism. Deposition of fibrillar amyloid proteins intraneuronally, as neurofibrillary tangles, extracellularly, as plaques and in blood vessels, is characteristic of both Alzheimer's disease and aged Down's syndrome. Amyloid deposition is also associated with type II diabetes mellitus.

Angiopoeitin

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Members of the angiopoeitin/fibrinogen family have been shown to stimulate the generation of new blood vessels, inhibit the generation of new blood vessels, and perform several roles in blood clotting. This generation of new blood vessels, called angiogenesis, is also an essential step in tumor growth in order for the tumor to get the blood supply it needs to expand. Variation in these genes may be predictive of any form of heart disease, numerous blood clotting disorders, stroke, hypertension and predisposition to tumor formation and metastasis. In particular, these variants may be predictive of the response to various antihypertensive drugs and chemotherapeutic and anti-tumor agents.

Apoptosis-related proteins

Active cell suicide (apoptosis) is induced by events such as growth factor withdrawal and toxins. It is controlled by regulators, which have either an inhibitory effect on programmed cell death (anti-apoptotic) or block the protective effect of inhibitors (proapoptotic). Many viruses have found a way of countering defensive apoptosis by encoding their own anti-apoptosis genes preventing their target-cells from dying too soon. Variants of apoptosis related genes may be useful in formulation of antiaging drugs.

Cadherin, Cyclin, Polymerase, Oncogenes, Histones, Kinases

Members of the cell division/cell cycle pathways such as cyclins, many transcription factors and kinases, DNA polymerases, histones, helicases and other oncogenes play a critical role in carcinogenesis where the uncontrolled proliferation of cells leads to tumor formation and eventually metastasis. Variation in these genes may be predictive of predisposition to any form of cancer, from increased risk of tumor formation to increased rate of metastasis. In particular, these variants may be predictive of the response to various chemotherapeutic and anti-tumor agents.

Colony-stimulating factor-related proteins

Granulocyte/macrophage colony-stimulating factors are cytokines that act in hematopoiesis by controlling the production, differentiation, and function of 2 related white cell populations of the blood, the granulocytes and the monocytes-macrophages.

5 Complement-related proteins

Complement proteins are immune associated cytotoxic agents, acting in a chain reaction to exterminate target cells to that were opsonized (primed) with antibodies, by forming a membrane attack complex (MAC). The mechanism of killing is by opening pores in the target cell membrane. Variations in 20 complement genes or their inhibitors are associated with many autoimmune disorders. Modified serum levels of complement products cause edemas of various tissues, lupus (SLE), vasculitis, glomerulonephritis, renal failure, hemolytic anemia, thrombocytopenia, and arthritis. They interfere with mechanisms of ADCC (antibody dependent cell cytotoxicity), severely impair immune competence and reduce phagocytic ability. Variants of complement genes may also be indicative of type I diabetes mellitus, meningitis neurological disorders such as Nemaline myopathy, Neonatal hypotonia, muscular disorders such as congenital myopathy and other diseases.

Cytochrome

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The respiratory chain is a key biochemical pathway which is essential to all aerobic cells. There are five different cytochromes involved in the chain. These are heme bound proteins which serve as electron carriers. Modifications in these genes may be predictive of ataxia areflexia, dementia and myopathic and neuropathic changes in muscles. Also, association with various types of solid tumors.

Kinesins

Kinesins are tubulin molecular motors that function to transport organelles within cells and to move chromosomes along microtubules during cell division. Modifications of these genes may be indicative of neurological disorders such as Pick disease of the brain, tuberous sclerosis.

Cytokines, Interferon, Interleukin

Members of the cytokine families are known for their potent ability to stimulate cell growth and division even at low concentrations. Cytokines such as erythropoietin are cell-specific in their growth stimulation; erythropoietin is useful for the stimulation of the proliferation of erythroblasts. Variants in cytokines may be predictive for a wide variety of diseases, including cancer predisposition.

G-protein coupled receptors

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G-protein coupled receptors (also called R7G) are an extensive group of hormones, neurotransmitters, odorants and light receptors which transduce extracellular signals by interaction with guanine nucleotide-binding (G) proteins. Alterations in genes coding for G-coupled proteins may be involved in and indicative of a vast number of physiological conditions. These include blood pressure regulation, renal dysfunctions, male infertility, dopamine associated cognitive, emotional, and endocrine functions, hypercalcemia, chondrodysplasia and osteoporosis, pseudohypoparathyroidism, growth retardation and dwarfism.

Thioesterases

Eukaryotic thiol proteases are a family of proteolytic enzymes which contain an active site cysteine. Catalysis proceeds through a thioester intermediate and is facilitated by a nearby histidine side chain; an asparagine completes the essential catalytic triad. Variants of thioester associated genes may be predictive of neuronal disorders and mental illnesses such as Ceroid Lipoffiscinosis, Neuronal 1, Infantile, Santavuori disease and more.

Breakdown Classifications of SNPS

The following list describes the numerical breakdown by molecule type of the SNPs described in Table 1. The key to these molecule types is as follows.

TPase_associated: 864
Guanylyl: 3
MHC: 1077
amylase: 44
30 amylaseinhib: 1
amyloid: 96
apoptosis: 91

WO 01/48245		PCT/US00/35346
	apoptosisinhib:	29
	apoptosisrecep:	14
	biotindep:	29
	cadhenn:	415
5	calcium channel:	85
	carboxylase:	4
	cathepsin:	336
	cathepsininhib:	41
	chloride_channel:	90
10	collagen:	1542
	complement:	222
	complementinhib:	21
	complementrecept:	10
	csf:	31
15	csf recept:	37
	cyclin:	65
	cyto45O:	136
	cytochrome:	659
	deaminase:	44
20	dehydrogenase:	1235
	desaturase:	9
	dna_rna_bind:	1309
	dna_rna_bind_inhib:	16
	dynein:	108
25	elastase:	134
	elastaseinhib:	6
	eph:	487
	esterase:	258
	esteraseinhib:	3
30 .	fgf:	34
	fgf receptor:	12
•	gaba:	45 .
	glucoamylase:	106
	glucuronidase:	14
35	glycoprotein:	3176
•	helicase:	333
	histone:	272
	homeobox:	431
	hydrolase:	187
40	hydroxysteroid:	84
	hypoxanthine:	4
	immunoglob:	1106
	immunoglob_recept:	19
	interferon:	322
45	interleukin:	88
	interleukinrecept:	126
	isomerase:	404
	isomeraseinhibitor:	45
	isomerasereceptor:	4
50	kinase:	1684

	kinase inhibitor:	187
	kinase receptor:	233
	kinesin:	86
	laminin:	196
5	lipase:	63
	metallothionein:	62
	misc_channel:	215
	ngf:	30
	nucl_recpt:	339
10	nuclease:	298
	oncogene:	783
	oxidase:	128
	oxygenase:	14
	peptidase:	150
15	peroxidase:	115
	phosphatase:	668
•	phosphataseinhib:	71
	phosphorylase:	84
	polymerase:	489
20	potassium_channel:	43
	prostaglandin:	55
	protease:	954
	proteaseinhib:	271
	reductase:	243
25	ribosomal prot:	1040
•	struct:	3128
	sulfotransferase:	42
	synthase:	893
	tgf:	117
30	tgfreceptor:	41
	thioesterase:	3
	thiolase:	38
	tm7:	453
	tnf:	151
35	tnfreceptor:	36
	traffic:	22
	transcriptfactor:	1139
	transferase:	291
	transport:	900
40	tubulin:	334
	ubiquitin:	229
	water_channel:	18
	unclassified:	10567

The key to the molecule type is as follows:

	Abbrev:	Title:
5	amylase amylaseinhib amyloid	amylase protein amylase inhibitor amyloid protein
10	apoptosis apoptosisinhib apoptosisrecep ATPase_associated	apoptosis associated protein apoptosis inhibitors apoptosis receptors ATPase associated protein
15	biotindep cadherin calcium_channel carboxylase	biotin dependent enzyme/protein cadherin protein calcium channel protein carboxylase protein
	cathepsin cathepsininhib chloride_channel collagen	cathepsin/carboxypeptidases cathepsin/carboxypeptidase inhibitor chloride channel protein collagen
20	complement complementrecept complementinhib csf	complement protein complement receptor protein complement inhibitor
25	csfrecept cyclin cyto450 cytochrome	colony stimulating factor colony stimulating factor receptor cyclin protein cytochrome p450 protein
30	deaminase dehydrogenase desaturase dna_rna_bind	cytochrome related protein deaminase dehydrogenase desaturase DNA/RNA binding protein/factor
35	dna_rna_inhib dynein elastase elastaseinhib	DNA/RNA binding protein/factor inhibitor dynein elastase elastase inhibitor
40	eph esterase esteraseinhib fgf	EPH family of tyrosine kinases esterase esterase inhibitor fibroblast growth factor
	fgfreceptor gaba glucoamylase glucoronidase	fibroblast growth factor receptor GABA receptor glucoamylase glucoronidase
45	glycoprotein Guanylyl helicase histone HOM	glycoprotein guanylylate cyclase helicase histone homologous

		1 C1/0300/33340
	homeobox	homeobox protein
	hydrolase	hydrolase
	hydroxysteroid	hydroxysteroid associated protein
	hypoxanthine	hypoxanthine associated protein
5	immunoglob	immunoglobulin
	immunoglobrecept	immunoglobulin receptor
	interferon	interferon
	interleukin	interleukin
	interleukinrecept	interleukin receptor
10	isomerase	isomerase
	isomeraseinhibitor	isomerase inhibitor
	isomerasereceptor	-
	kinase	isomerase receptor kinase
	kinaseinhibitor	
15		kinase inhibitor
13	kinasereceptor kinesin	kinase receptor
		kinesin
	laminin	laminin associated protein
	lipase	lipase
22	metallothionein	metallothionein
20	MHC	major histocompatability complex
	misc_channel	miscellaneous channel
	ngf	nerve growth factor
	nuci_recpt	nuclear receptor
	nuclease	nuclease
25	oncogene	oncogene associated protein
	oxidase	oxidase
	oxygenase	oxygenase
	peptidase	peptidase
	peroxidase	peroxidase
30	phosphatase	phosphatase
	phosphataseinhib	phosphatase inhibitor
	phosphorylase	phosphorylase
	PIR	PIR DATABASE (release 56, 29-OCT-
		1998)
35	polymerase	polymerase
	potassium_channel	potassium channel protein
	prostaglandin	prostaglandin
	protease	protease
	proteaseinhib	protease inhibitor
40	reductase	reductase
	ribosomalprot	ribosomal associated protein
	RTR	EMBLDATABASE translated entries
		not to be incorporated into SWISS-
		PROT (20-JUL-1998)
45	SIM	similar
10	SPTR	EMBL DATABASE translated entries to
	21 III	
		be incorporated into SWISS-PROT (20-
	struct	JUL-1998)
50	sulfotransferase	structural associated protein
50	201101101101020	sulfotransferase

	WO 01/48245	PCT/US00/35346
	SWP	SWISS-PROT DATABASE (release 18- OCT-1998)
	SWPN	SWISS-PROT Update (release 11-NOV-98)
5	synthase	synthase
	tgf	transforming growth factor
	tgfreceptor	transforming growth factor receptor
	thioesterase	thioesterase
	thiolase	thiolase
10	tm7	seven transmembrane domain G-protein coupled receptor
	tnf	necrosis factor receptor
	traffic	tumor necrosis factor
	tnfreceptor	tumor trafficking associated protein
15	TRN	EMBL DATABASE translated entries update (20-JUL-1998)
	transcriptfactor	transcription factor
	transferase	transferase
	transport	transport protein
20	tubulin	tubulin
	ubiquitin	ubiquitin
	unclassified	Protein not categorized into one of the aforementioned protein families
	water channel	water channel protein

Table 1

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A compilation of polymorphisms is listed in Table 1. Table 1 includes thirteen columns that provide descriptive information for each cSNP, each of which occupies one row in the Table. The column headings, and an explanation for each, are given below.

The first column of the table lists the names assigned to the fragments in which the polymorphisms occur. The fragments are all human genomic fragments. The sequence of one allelic form of each of the fragments (arbitrarily referred to as the prototypical or reference form) has been previously published. These sequences are listed at http://www-genome.wi.mit.edu/ (all STS's sequence tag sites)); http://shgc.stanford.edu (Stanford STS's); and http://www.tigr.org/ (TIGR STS's). The web sites also list primers for amplification of the fragments, and the genomic location of the fragments. Some fragments are expressed sequence tags, and some are random genomic fragments. All information in the web sites concerning the fragments listed in the table is incorporated by reference in its entirety for all purposes.

The second column lists the position in the fragment in which a polymorphic site has been found. Positions are numbered consecutively with the first base of the fragment sequence listed as in one of the above databases being assigned the number one. The third column lists the base occupying the polymorphic site in the sequence in the data base. This base is arbitrarily designated the reference or prototypical form, but it is not necessarily the most frequently occurring form. The fourth column in the table lists the alternative base(s) at the polymorphic site. The fifth column of the table lists a 5' (upstream or forward) primer that hybridizes with the 5' end of the DNA sequence to be amplified. The sixth column of the table lists a 3' (downstream or reverse) primer that hybridizes with the complement of the 3' end of the sequence to be amplified. The seventh column of the table lists a number of bases of sequence on either side of the polymorphic site in each fragment. The indicated sequences can either be DNA or RNA. In the latter, the T's shown in the table are replaced by U's. The base occupying the polymorphic site is indicated in EUT'AC-IUB ambiguity code.

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"SEQ ID" provides the cross-references to the two nucleotide SEQ ID NOS: for the cognate pair, which are numbered consecutively, and, as explained below, amino acid SEQ ID NOS: as well, in the Sequence Listing of the application.

Each sequence entry in the Sequence Listing also includes a cross-reference to the CuraGen sequence ID, under the label "Accession number". The first pair of SEQ ID NOS: given in the first column of each row of the Table is the SEQ ID NO: identifying the nucleic acid sequence for the polymorphism. If a polymorphism carries an entry for the amino acid portion of the row, a third SEQ ID NO: appears in parentheses in the column "Amino acid before" (see below) for the reference amino acid sequence, and a fourth SEQ ID NO: appears in parentheses in the column "Amino acid after" (see below) for the polymorphic amino acid sequence. The latter SEQ ID NOS: refer to amino acid sequences giving the cognate reference and polymorphic amino acid sequences that are the translation of the nucleotide polymorphism. If a polymorphism carries no entry for the protein portion of the row, only one pair SEQ ID NOS: is provided, in the first column.

"CuraGen sequence ID" provides CuraGen Corporation's accession number.

"Base pos. of SNP" gives the numerical position of the nucleotide in the nucleic acid at which the cSNP is found, as identified in this invention.

"Polymorphic sequence" provides a 51-base sequence with the polymorphic site at the 26th base in the sequence, as well as 25 bases from the reference sequence on the 5' side and the 3' side of the polymorphic site. The designation at the polymorphic site is enclosed in square brackets, and provides first, the reference nucleotide; second, a "slash (/)"; and third, the polymorphic nucleotide. In certain cases the polymorphism is an insertion or a deletion. In that case, the position that is "unfilled" (i.e., the reference or the polymorphic position) is indicated by the word "gap".

"Base before" provides the nucleotide present in the reference sequence at the position at which the polymorphism is found.

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"Base after" provides the altered nucleotide at the position of the polymorphism.

"Amino acid before" provides the amino acid in the reference protein, if the polymorphism occurs in a coding region. This column also includes the SEQ ID NO: in parentheses for the translated reference amino acid sequence if the polymorphism occurs in a coding region.

"Amino acid after" provides the amino acid in the polymorphic protein, if the polymorphism occurs in a coding region. This column also includes the SEQ ID NO in parentheses for the translated polymorphic amino acid sequence if the polymorphism occurs in a coding region.

"Type of change" provides information on the nature of the polymorphism.

"SILENT-NONCODING" is used if the polymorphism occurs in a noncoding region of a nucleic acid. "SILENT-CODING" is used if the polymorphism occurs in a coding region of a nucleic acid of a nucleic acid and results in no change of amino acid in the translated polymorphic protein. "CONSERVATIVE" is used if the polymorphism occurs in a coding region of a nucleic acid and provides a change in which the altered amino acid falls in the same class as the reference amino acid. The classes are: 1) Aliphatic: Gly, Ala, Val, Leu, Ile; 2) Aromatic: Phe, Tyr, Trp; 3) Sulfur-containing: Cys, Met; 4) Aliphatic OH: Ser, Thr; 5) Basic: Lys, Arg, His; 6) Acidic: Asp, Glu, Asn, Gln; 7) Pro falls in none of the other classes; and 8) End defines a termination codon.

"NONCONSERVATIVE" is used if the polymorphism occurs in a coding region of a nucleic acid and provides a change in which the altered amino acid falls in a different class than the reference amino acid.

"FRAMESHIFT" relates to an insertion or a deletion. If the frameshift occurs in a coding region, the Table provides the translation of the frameshifted codons 3' to the polymorphic site.

"Protein classification of CuraGen gene" provides a generic class into which the protein is classified. Multiple classes of proteins were identified as listed above in the discussion of Table 1.

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"Name of protein identified following a BLASTX analysis of the CuraGen sequence" provides the database reference for the protein found to resemble the novel reference-polymorphism cognate pair most closely.

"Similarity (pvalue) following a BLASTX analysis" provides the pvalue, a statistical measure from the BLASTX analysis that the polymorphic sequence is similar to, and therefore an allele of, the reference, or wild-type, sequence. In the present application, a cutoff of pvalue $> 1 \times 10^{-50}$ (entered, for example, as 1.0E-50 in the Table) is used to establish that the reference-polymorphic cognate pairs are novel. A pvalue $< 1 \times 10^{-50}$ defines proteins considered to be already known.

"Map location" provides any information available at the time of filing related to localization of a gene on a chromosome.

The polymorphisms are arranged in Table 1 in the following order:

SEQ ID NOs: 1-422 are nucleotide sequences for SNPs that are silent.

SEQ ID NOs: 423-480 are nucleotide sequences for SNPs that lead to conservative amino acid changes.

SEQ ID NOs: 481-619 are nucleotide sequences for SNPs that lead to nonconservative amino acid changes.

SEQ ID NOs: 620-651 are nucleotide sequences for SNPs that involve a gap. With respect to the reference or wild-type sequence at the position of the polymorphism, the allelic

cSNP introduces an additional nucleotide (an insertion) or deletes a nucleotide (a deletion). An SNP that involves a gap generates a frame shift.

Also presented in the sequence listing filed herewith are predicted amino acid sequences encoded by the polymorphic sequences shown in Table 1.

SEQ ID NOs: 652-709 are the amino acid sequences centered at the polymorphic amino acid residue for the protein products provided by SNPs that lead to conservative amino acid changes. 7 or 8 amino acids on either side of the polymorphic site are shown. The order in which these sequences appear mirrors the order of presentation of the cognate nucleotide sequences, and is set forth in the Table.

SEQ ID NOs: 710-848 are the amino acid sequences centered at the polymorphic amino acid residue for the protein products provided by SNPs that lead to nonconservative amino acid changes. 7 or 8 amino acids on either side of the polymorphic site are shown. The order in which these sequences appear mirrors the order of presentation of the cognate nucleotide sequences, and is set forth in the Table.

SEQ ID NOs: 849-880 are the amino acid sequences centered at the polymorphic amino acid residue for the protein products provided by SNPs that lead to frameshift-induced amino acid changes. 7 or 8 amino acids on either side of the polymorphic site are shown. The order in which these sequences appear mirrors the order of presentation of the cognate nucleotide sequences, and is set forth in the Table.

Provided herein are compositions which include, or are capable of detecting, nucleic acid sequences having these polymorphisms, as well as methods of using nucleic acids.

Identification of Individuals Carrying SNPs

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Individuals carrying polymorphic alleles of the invention may be detected at either the DNA, the RNA, or the protein level using a variety of techniques that are well known in the art. Strategies for identification and detection are described in e.g., EP 730,663, EP 717,113, and PCT US97/02102. The present methods usually employ pre-characterized polymorphisms. That is, the genotyping location and nature of polymorphic forms present at a site have already been determined. The availability of this information allows sets of probes to be designed for specific identification of the known polymorphic forms.

Many of the methods described below require amplification of DNA from target samples. This can be accomplished by e.g., PCR. (1989), B. for detecting polymorphisms. See generally PCR Technology: Principles and Applications for DNA Amplification (ed. H.A. Erlich, Freeman Press, NY, NY, 1992); PCR Protocols: A Guide to Methods and Applications (eds. Innis, et al., Academic Press, San Diego, CA, 1990); Mattila et al., Nucleic Acids Res. 19, 4967 (1991); Eckert et al., PCR Methods and Applications 1, 17 (1991); PCR (eds. McPherson et al., IRL Press, Oxford); and U.S. Patent 4,683,202.

The phrase "recombinant protein" or "recombinantly produced protein" refers to a peptide or protein produced using non-native cells that do not have an endogenous copy of DNA able to express the protein. In particular, as used herein, a recombinantly produced protein relates to the gene product of a polymorphic allele, i.e., a "polymorphic protein" containing an altered amino acid at the site of translation of the nucleotide polymorphism. The cells produce the protein because they have been genetically altered by the introduction of the appropriate nucleic acid sequence. The recombinant protein will not be found in association with proteins and other subcellular components normally associated with the cells producing the protein. The terms "protein" and "polypeptide" are used interchangeably herein.

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The phrase "substantially purified" or "isolated" when referring to a nucleic acid, peptide or protein, means that the chemical composition is in a milieu containing fewer, or preferably, essentially none, of other cellular components with which it is naturally associated. Thus, the phrase "isolated" or "substantially pure" refers to nucleic acid preparations that lack at least one protein or nucleic acid normally associated with the nucleic acid in a host cell. It is preferably in a homogeneous state although it can be in either a dry or aqueous solution. Purity and homogeneity are typically determined using analytical chemistry techniques such as gel electrophoresis or high performance liquid chromatography. Generally, a substantially purified or isolated nucleic acid or protein will comprise more than 80% of all macromolecular species present in the preparation. Preferably, the nucleic acid or protein is purified to represent greater than 90% of all macromolecular species present. More preferably the nucleic acid or protein is purified to greater than 95%, and most preferably the nucleic acid or protein is purified to essential homogeneity, wherein other macromolecular species are not detected by conventional analytical procedures.

The genomic DNA used for the diagnosis may be obtained from any nucleated cells of the body, such as those present in peripheral blood, urine, saliva, buccal samples, surgical specimen, and autopsy specimens. The DNA may be used directly or may be amplified enzymatically in vitro through use of PCR (Saiki et al. Science 239:487-491 (1988)) or other in vitro amplification methods such as the ligase chain reaction (LCR) (Wu and Wallace Genomics 4:560-569 (1989)), strand displacement amplification (SDA) (Walker et al. Proc. Natl. Acad. Sci. U.S.A. 89:392-396 (1992)), self-sustained sequence replication (3SR) (Fahy et al. PCR Methods P&J& 1:25-33 (1992)), prior to mutation analysis.

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The method for preparing nucleic acids in a form that is suitable for mutation detection is well known in the art. A "nucleic acid" is a deoxyribonucleotide or ribonucleotide polymer in either single-or double-stranded form, including known analogs of natural nucleotides unless otherwise indicated. The term "nucleic acids", as used herein, refers to either DNA or RNA. "Nucleic acid sequence" or "polynucleotide sequence" refers to a single-stranded sequence of deoxyribonucleotide or ribonucleotide bases read from the 5' end to the 3' end. The direction of 5' to 3' addition of nascent RNA transcripts is referred to as the transcription direction; sequence regions on the DNA strand having the same sequence as the RNA and which are beyond the 5' end of the RNA transcript in the 5' direction are referred to as "upstream sequences"; sequence regions on the DNA strand having the same sequence as the RNA and which are beyond the 3' end of the RNA transcript in the 3' direction are referred to as "downstream sequences". The term includes both self-replicating plasmids, infectious polymers of DNA or RNA and nonfunctional DNA or RNA. The complement of any nucleic acid sequence of the invention is understood to be included in the definition of that sequence. "Nucleic acid probes" may be DNA or RNA fragments.

The detection of polymorphisms in specific DNA sequences, can be accomplished by a variety of methods including, but not limited to, restriction-fragment-length-polymorphism detection based on allele-specific restriction-endonuclease cleavage (Kan and Dozy Lancet ii:910-912 (1978)), hybridization with allele-specific oligonucleotide probes (Wallace et al. Nucl. Acids Res. 6:3543-3557 (1978)), including immobilized oligonucleotides (Saiki et al. Proc. Natl. Acad. SCI. USA, 86:6230-6234 (1969)) or oligonucleotide arrays (Maskos and Southern Nucl. Acids Res. 21:2269-2270 (1993)), allele-specific PCR (Newton et al. Nucl. Acids Res. 17:2503-2516 (1989)), mismatch-repair detection (MRD) (Faham and Cox. Genome Res. 5:474-482 (1995)), binding of MutS protein (Wagner et al. Nucl. Acids Res. 23:3944-3948 (1995), denaturing-gradient gel electrophoresis (DGGE) (Fisher and Lerman et

al. *Proc. Natl. Acad. Sci. U.S.A. 80:1579-1* 583 (1983)), single-strand-conformation-polymorphism detection (Orita et al. Genomics 5:874-879 (1983)), RNAase cleavage at mismatched base-pairs (Myers et al. Science 230:1242 (1985)), chemical (Cotton et al. Proc. Natl. w Sci. U.S.A, 8Z4397-4401 (1988)) or enzymatic (Youil et al. Proc. Natl. Acad. Sci. U.S.A. 92:87-91 (1995)) cleavage of heteroduplex DNA, methods based on allele specific primer extension (Syvanen et al. Genomics 8:684-692 (1990)), genetic bit analysis (GBA) (Nikiforov et al. &&I Acids 22:4167-4175 (1994)), the oligonucleotide-ligation assay (OLA) (Landegren et al. Science 241:1077 (1988)), the allele-specific ligation chain reaction (LCR) (Barrany Proc. Natl. Acad. Sci. U.S.A. 88:189-1 93 (1991)), gap-LCR (Abravaya et al. Nucl Acids Res 23:675-682 (1995)), radioactive and/or fluorescent DNA sequencing using standard procedures well known in the art, and peptide nucleic acid (PNA) assays (Orum et al., Nucl. Acids Res, 21:5332-5356 (1993); Thiede et al., Nucl. Acids Res. 24:983-984 (1996)).

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"Specific hybridization" or "selective hybridization" refers to the binding, or duplexing, of a nucleic acid molecule only to a second particular nucleotide sequence to which the nucleic acid is complementary, under suitably stringent conditions when that sequence is present in a complex mixture (e.g., total cellular DNA or RNA). "Stringent conditions" are conditions under which a probe will hybridize to its target subsequence, but to no other sequences. Stringent conditions are sequence-dependent and are different in different circumstances. Longer sequences hybridize specifically at higher temperatures than shorter ones. Generally, stringent conditions are selected such that the temperature is about 5°C lower than the thermal melting point (Tm) for the specific sequence to which hybridization is intended to occur at a defined ionic strength and pH. The Tm is the temperature (under defined ionic strength, pH, and nucleic acid concentration) at which 50% of the target sequence hybridizes to the complementary probe at equilibrium. Typically, stringent conditions include a salt concentration of at least about 0.01 to about 1.0 M Na ion concentration (or other salts), at pH 7.0 to 8.3. The temperature is at least about 30°C for short probes (e.g., 10 to 50 nucleotides). Stringent conditions can also be achieved with the addition of destabilizing agents such as formamide. For example, conditions of 5X SSPE (750 mM NaCl, 50 mM NaPhosphate, 5 mM EDTA, pH 7.4) and a temperature of 25-30°C are suitable for allele-specific probe hybridizations.

"Complementary" or "target" nucleic acid sequences refer to those nucleic acid sequences which selectively hybridize to a nucleic acid probe. Proper annealing conditions

depend, for example, upon a probe's length, base composition, and the number of mismatches and their position on the probe, and must often be determined empirically. For discussions of nucleic acid probe design and annealing conditions, see, for example, Sambrook et al., or <u>Current Protocols in Molecular Biology</u>, F. Ausubel *et al.*, ed., Greene Publishing and Wiley-Interscience, New York (1987).

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A perfectly matched probe has a sequence perfectly complementary to a particular target sequence. The test probe is typically perfectly complementary to a portion of the target sequence. A "polymorphic" marker or site is the locus at which a sequence difference occurs with respect to a reference sequence. Polymorphic markers include restriction fragment length polymorphisms, variable number of tandem repeats (VNTR's), hypervariable regions, minisatellites, dinucleotide repeats, trinucleotide repeats, tetranucleotide repeats, simple sequence repeats, and insertion elements such as Alu. The reference allelic form may be, for example, the most abundant form in a population, or the first allelic form to be identified, and other allelic forms are designated as alternative, variant or polymorphic alleles. The allelic form occurring most frequently in a selected population is sometimes referred to as the "wild type" form, and herein may also be referred to as the "reference" form. Diploid organisms may be homozygous or heterozygous for allelic forms. A diallelic polymorphism has two distinguishable forms (i.e., base sequences), and a triallelic polymorphism has three such forms.

As use herein an "oligonucleotide" is a single-stranded nucleic acid ranging in length from 2 to about 60 bases. Oligonucleotides are often synthetic but can also be produced from naturally occurring polynucleotides. A probe is an oligonucleotide capable of binding to a target nucleic acid of complementary sequence through one or more types of chemical bonds, usually through complementary base pairing via hydrogen bond formation. Oligonucleotides probes are often between 5 and 60 bases, and, in specific embodiments, may be between 10-40, or 15-30 bases long. An oligonucleotide probe may include natural (i.e. A, G, C, or T) or modified bases (7-deazaguanosine, inosine, etc.). In addition, the bases in an oligonucleotide probe may be joined by a linkage other than a phosphodiester bond, such as a phosphoramidite linkage or a phosphorothioate linkage, or they may be peptide nucleic acids in which the constituent bases are joined by peptide bonds rather than by phosphodiester bonds, so long as it does not interfere with hybridization. Examples of an oligonucleotide are shown in Table 1. Oligonucleotides can be all of a nucleic acid segment as represented in column 4 of Table 1; a nucleic acid sequence which comprises a nucleic acid segment

represented in column 4 of Table 1 and additional nucleic acids (present at either or both ends of a nucleic acid segment of column 4); or a portion (fragment) of a nucleic acid segment represented in column 4 of the table which includes a polymorphic site. Preferred polymorphic sites of the invention include segments of DNA or their complements, which include any one of the polymorphic sites shown in the Table. The segments can be between 5 and 250 bases, and, in specific embodiments are between 5-10, 5-20, 10-20, 10-50, 20-50 or 10-100 bases. The polymorphic site can occur within any position of the segment. The segments can be from any of the allelic forms of the DNA shown in the Table.

As used herein, the term "primer" refers to a single-stranded oligonucleotide which acts as a point of initiation of template-directed DNA synthesis under appropriate conditions (e.g., in the presence of four different nucleoside triphosphates and a polymerization agent, such as DNA polymerase, RNA polymerase or reverse transcriptase) in an appropriate buffer and at a suitable temperature. The appropriate length of a primer depends on the intended use of the primer, but typically ranges from 15 to 30 nucleotides. Short primer molecules generally require cooler temperatures to form sufficiently stable hybrid complexes with the template. A primer need not be perfectly complementary to the exact sequence of the template, but should be sufficiently complementary to hybridize with it. The term "primer site" refers to the sequence of the target DNA to which a primer hybridizes. The term "primer pair" refers to a set of primers including a 5' (upstream) primer that hybridizes with the 5' end of the DNA sequence to be amplified and a 3' (downstream) primer that hybridizes with the complement of the 3' end of the sequence to be amplified.

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DNA fragments can be prepared, for example, by digesting plasmid DNA, or by use of PCR. Oligonucleotides for use as primers or probes are chemically synthesized by methods known in the field of the chemical synthesis of polynucleotides, including by way of non-limiting example the phosphoramidite method described by Beaucage and Carruthers, Tetrahedron Lett 22:1859-1862 (1981) and the triester method provided by Matteucci, et al., J. Am. Chem. Soc., 103:3185 (1981) both incorporated herein by reference. These syntheses may employ an automated synthesizer, as described in Needham-VanDevanter, D.R., et al., Nucleic Acids Res. 12:61596168 (1984). Purification of oligonucleotides may be carried out by either native acrylamide gel electrophoresis or by anion-exchange HPLC as described in Pearson, J.D. and Regnier, F.E., ,J. Chrom., 255:137-149 (1983). A double stranded fragment may then be obtained, if desired, by annealing appropriate complementary single strands together under suitable conditions or by synthesizing the complementary strand

using a DNA polymerase with an appropriate primer sequence. Where a specific sequence for a nucleic acid probe is given, it is understood that the complementary strand is also identified and included. The complementary strand will work equally well in situations where the target is a double-stranded nucleic acid.

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The sequence of the synthetic oligonucleotide or of any nucleic acid fragment can be can be obtained using either the dideoxy chain termination method or the Maxam-Gilbert method (see Sambrook et al. Molecular Cloning - a Laboratory Manual (2nd Ed.), Vols. 1-3, Cold Spring Harbor Laboratory, Cold Spring Harbor, New York, (1989), which is incorporated herein by reference. This manual is hereinafter referred to as "Sambrook et al." ______; Zyskind et al., (1988)). Recombinant DNA Laboratory Manual, (Acad. Press, New York). Oligonucleotides useful in diagnostic assays are typically at least 8 consecutive nucleotides in length, and may range upwards of 18 nucleotides in length to greater than 100 or more consecutive nucleotides.

Another aspect of the invention pertains to isolated antisense nucleic acid molecules that are hybridizable to or complementary to the nucleic acid molecule comprising the SNP-containing nucleotide sequences of the invention, or fragments, analogs or derivatives thereof. An "antisense" nucleic acid comprises a nucleotide sequence that is complementary to a "sense" nucleic acid encoding a protein, e.g., complementary to the coding strand of a double-stranded cDNA molecule or complementary to an mRNA sequence. In specific aspects, antisense nucleic acid molecules are provided that comprise a sequence complementary to at least about 10, about 25, about 50, or about 60 nucleotides or an entire SNP coding strand, or to only a portion thereof.

In one embodiment, an antisense nucleic acid molecule is antisense to a "coding region" of the coding strand of a polymorphic nucleotide sequence of the invention. The term "coding region" refers to the region of the nucleotide sequence comprising codons which are translated into amino acid. In another embodiment, the antisense nucleic acid molecule is antisense to a "noncoding region" of the coding strand of a nucleotide sequence of the invention. The term "noncoding region" refers to 5' and 3' sequences which flank the coding region that are not translated into amino acids (*i.e.*, also referred to as 5' and 3' untranslated regions).

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Given the coding strand sequences disclosed herein, antisense nucleic acids of the invention can be designed according to the rules of Watson and Crick or Hoogsteen base pairing. For example, the antisense nucleic acid molecule can generally be complementary to the entire coding region of an mRNA, but more preferably as embodied herein, it is an oligonucleotide that is antisense to only a portion of the coding or noncoding region of the mRNA. An antisense oligonucleotide can range in length between about 5 and about 60 nucleotides, preferably between about 10 and about 45 nucleotides, more preferably between about 15 and 30 in length. An antisense nucleic acid of the invention can be constructed using chemical synthesis or enzymatic ligation reactions using procedures known in the art. For example, an antisense nucleic acid (e.g., an antisense oligonucleotide) can be chemically synthesized using naturally occurring nucleotides or variously modified nucleotides designed to increase the biological stability of the molecules or to increase the physical stability of the duplex formed between the antisense and sense nucleic acids, e.g., phosphorothioate derivatives and acridine substituted nucleotides can be used.

Examples of modified nucleotides that can be used to generate the antisense nucleic acid include: 5-fluorouracil, 5-bromouracil, 5-chlorouracil, 5-iodouracil, hypoxanthine, xanthine, 4-acetylcytosine, 5-(carboxyhydroxylmethyl) uracil, 5-carboxymethylaminomethyl-2-thiouridine, 5-carboxymethylaminomethyluracil, dihydrouracil, beta-D-galactosylqueosine, inosine, N6-isopentenyladenine, 1-methylguanine, 1-methylinosine, 2,2-dimethylguanine, 2-methyladenine, 2-methylguanine, 3-methylcytosine, 5-methylcytosine, N6-adenine, 7-methylguanine, 5-methylaminomethyluracil, 5-methoxyaminomethyl-2-thiouracil, beta-D-mannosylqueosine, 5'-methoxycarboxymethyluracil, 5-methoxyuracil, 2-methylthio-N6-isopentenyladenine, uracil-5-oxyacetic acid (v), wybutoxosine, pseudouracil, queosine, 2-thiocytosine, 5-methyl-2-thiouracil, 2-thiouracil, 4-thiouracil, 5-methyluracil, uracil-5-oxyacetic acid methylester, uracil-5-oxyacetic acid (v), 5-methyl-2-thiouracil, 3-(3-amino-3-N-2-carboxypropyl) uracil, (acp3)w, and 2,6-diaminopurine. Alternatively, the antisense nucleic acid can be produced biologically using an expression vector into which a nucleic acid has been subcloned in an antisense orientation (i.e., RNA transcribed from the inserted nucleic acid will be of an antisense orientation to a target nucleic acid of interest, described further in the following subsection).

The antisense nucleic acid molecules of the invention are typically administered to a subject or generated *in situ* such that they hybridize with or bind to cellular mRNA and/or

genomic DNA encoding a polymorphic protein to thereby inhibit expression of the protein, e.g., by inhibiting transcription and/or translation. The hybridization can be by conventional nucleotide complementary to form a stable duplex, or, for example, in the case of an antisense nucleic acid molecule that binds to DNA duplexes, through specific interactions in the major groove of the double helix. An example of a route of administration of antisense nucleic acid molecules of the invention includes direct injection at a tissue site.

Alternatively, antisense nucleic acid molecules can be modified to target selected cells and then administered systemically. For example, for systemic administration, antisense molecules can be modified such that they specifically bind to receptors or antigens expressed on a selected cell surface, e.g., by linking the antisense nucleic acid molecules to peptides or antibodies that bind to cell surface receptors or antigens. The antisense nucleic acid molecules can also be delivered to cells using the vectors described herein. To achieve sufficient intracellular concentrations of antisense molecules, vector constructs in which the antisense nucleic acid molecule is placed under the control of a strong pol II or pol III promoter are preferred.

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In yet another embodiment, the antisense nucleic acid molecule of the invention is an α-anomeric nucleic acid molecule. An α-anomeric nucleic acid molecule forms specific double-stranded hybrids with complementary RNA in which, contrary to the usual -units, the strands run parallel to each other (Gaultier et al. (1987) Nucleic Acids Res 15: 6625-6641). The antisense nucleic acid molecule can also comprise a 2'-o-methylribonucleotide (Inoue et al. (1987) Nucleic Acids Res 15: 6131-6148) or a chimeric RNA -DNA analogue (Inoue et al. (1987) FEBS Lett 215: 327-330).

The following terms are used to describe the sequence relationships between two or more nucleic acids or polynucleotides: "reference sequence", "comparison window", "sequence identity", "percentage of sequence identity", and "substantial identity". A "reference sequence" is a defined sequence used as a basis for a sequence comparison; a reference sequence may be a subset of a larger sequence, for example, as a segment of a full-length cDNA or gene sequence given in a sequence listing, or may comprise a complete cDNA or gene sequence. Optimal alignment of sequences for aligning a comparison window may, for example, be conducted by the local homology algorithm of Smith and Waterman Adv. Appl. Math. 2482 (1981), by the homology alignment algorithm of Needleman and Wunsch J. Mol. Biol. 48:443 (1970), by the search for similarity method of Pearson and

Lipman <u>Proc. Natl. Acad. Sci. U.S.A.</u> 852444 (1988), or by computerized implementations of these algorithms (for example, GAP, BESTFIT, FASTA, and TFASTA in the Wisconsin Genetics Software Package Release 7.0, Genetics Computer Group, 575 Science Dr., Madison, WI).

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Techniques for nucleic acid manipulation of the nucleic acid sequences harboring the cSNP's of the invention, such as subcloning nucleic acid sequences encoding polypeptides into expression vectors, labeling probes, DNA hybridization, and the like, are described generally in Sambrook et al., The phrase "nucleic acid sequence encoding" refers to a nucleic acid which directs the expression of a specific protein, peptide or amino acid sequence. The nucleic acid sequences include both the DNA strand sequence that is transcribed into RNA and the RNA sequence that is translated into protein, peptide or amino acid sequence. The nucleic acid sequences include both the full length nucleic acid sequences disclosed herein as well as non-full length sequences derived from the full length protein. It being further understood that the sequence includes the degenerate codons of the native sequence or sequences which may be introduced to provide codon preference in a specific host cell. Consequently, the principles of probe selection and array design can readily be extended to analyze more complex polymorphisms (see EP 730,663). For example, to characterize a triallelic SNP polymorphism, three groups of probes can be designed tiled on the three polymorphic forms as described above. As a further example, to analyze a diallelic polymorphism involving a deletion of a nucleotide, one can tile a first group of probes based on the undeleted polymorphic form as the reference sequence and a second group of probes based on the deleted form as the reference sequence.

For assay of genomic DNA, virtually any biological convenient tissue samples include whole blood, semen, saliva, tears, urine, fecal material, sweat, buccal, skin and hair can be used. Genomic DNA is typically amplified before analysis. Amplification is usually effected by PCR using primers flanking a suitable fragment e.g., of 50-500 nucleotides containing the locus of the polymorphism to be analyzed. Target is usually labeled in the course of amplification. The amplification product can be RNA or DNA, single stranded or double stranded. If double stranded, the amplification product is typically denatured before application to an array. If genomic DNA is analyzed without amplification, it may be desirable to remove RNA from the sample before applying it to the array. Such can be accomplished by digestion with DNase-free RNAase.

DETECTION OF POLYMORPHISMS IN A NUCLEIC ACID SAMPLE

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The SNPs disclosed herein can be used to determine which forms of a characterized polymorphism are present in individuals under analysis.

The design and use of allele-specific probes for analyzing polymorphisms is described by e.g., Saiki et al., Nature 324, 163-166 (1986); Dattagupta, EP 235,726, Saiki, WO 89/11548. Allele-specific probes can be designed that hybridize to a segment of target DNA from one individual but do not hybridize to the corresponding segment from another individual due to the presence of different polymorphic forms in the respective segments from the two individuals. Hybridization conditions should be sufficiently stringent that there is a significant difference in hybridization intensity between alleles, and preferably an essentially binary response, whereby a probe hybridizes to only one of the alleles. Some probes are designed to hybridize to a segment of target DNA such that the polymorphic site aligns with a central position (e.g., in a 15-mer at the 7 position; in a 16-mer, at either the 7, 8 or 9 position) of the probe. This design of probe achieves good discrimination in hybridization between different allelic forms.

Allele-specific probes are often used in pairs, one member of a pair showing a perfect match to a reference form of a target sequence and the other member showing a perfect match to a variant form. Several pairs of probes can then be immobilized on the same support for simultaneous analysis of multiple polymorphisms within the same target sequence.

The polymorphisms can also be identified by hybridization to nucleic acid arrays, some examples of which are described in oublished PCT application WO 95/11995. WO 95/11995 also describes subarrays that are optimized for detection of a variant form of a precharacterized polymorphism. Such a subarray contains probes designed to be complementary to a second reference sequence, which is an allelic variant of the first reference sequence. The second group of probes is designed by the same principles, except that the probes exhibit complementarity to the second reference sequence. The inclusion of a second group (or further groups) can be particularly useful for analyzing short subsequences of the primary reference sequence in which multiple mutations are expected to occur within a short distance commensurate with the length of the probes (e.g., two or more mutations within 9 to 21 bases).

An allele-specific primer hybridizes to a site on target DNA overlapping a polymorphism and only primes amplification of an allelic form to which the primer exhibits perfect complementarity. See Gibbs, Nucleic Acid Res. 17 2427-2448 (1989). This primer is used in conjunction with a second primer which hybridizes at a distal site. Amplification proceeds from the two-primers, resulting in a detectable product which indicates the particular allelic form is present. A control is usually performed with a second pair of primers, one of which shows a single base mismatch at the polymorphic site and the other of which exhibits perfect complementarity to a distal site. The single-base mismatch prevents amplification and no detectable product is formed. The method works best when the mismatch is included in the 3'-most position of the oligonucleotide aligned with the polymorphism because this position is most destabilizing to elongation from the primer (see, e.g., WO 93/22456).

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Amplification products generated using the polymerase chain reaction can be analyzed by the use of denaturing gradient gel electrophoresis. Different alleles can be identified based on the different sequence-dependent melting properties and electrophoretic migration of DNA in solution. Erlich, ed., PCR Technology, Principles and Applications, for DNA Amplification, (W.H. Freeman and Co New York, 1992, Chapter 7).

Alleles of target sequences can be differentiated using single-strand conformation polymorphism analysis, which identifies base differences by alteration in electrophoretic migration of single stranded PCR products, as described in Orita et al., Proc. Nat. Acad. Sci. 86, 2766-2770 (1989). Amplified PCR products can be generated and heated or otherwise denatured, to form single stranded amplification products. Single-stranded nucleic acids may refold or form secondary structures which are partially dependent on the base sequence. The different electrophoretic mobilities of single-stranded amplification products can be related to base-sequence differences between alleles of target sequences.

The genotype of an individual with respect to a pathology suspected of being caused by a genetic polymorphism may be assessed by association analysis. Phenotypic traits suitable for association analysis include diseases that have known but hitherto unmapped genetic components (e.g., agammaglobulinemia, diabetes insipidus, Lesch-Nyhan syndrome, muscular dystrophy, Wiskott-Aldrich syndrome, Fabry's disease, familial hypercholesterolemia, polycystic kidney disease, hereditary spherocytosis, von Willebrand's

disease, tuberous sclerosis, hereditary hemorrhagic telangiectasia, familial colonic polyposis, Ehlers-Danlos syndrome, osteogenesis imperfecta, and acute intermittent porphyria).

Phenotypic traits also include symptoms of, or susceptibility to, multifactorial diseases of which a component is or may be genetic, such as autoimmune diseases, inflammation, cancer, system, diseases of the nervous and infection by pathogenic microorganisms. Some examples of autoimmune diseases include rheumatoid arthritis, multiple sclerosis, diabetes (insulin-dependent and non- independent), systemic lupus erythematosus and Graves disease. Some examples of cancers include cancers of the bladder, brain, breast, colon, esophagus, kidney, oral cavity, ovary, pancreas, prostate, skin, stomach, leukemia, liver, lung, and uterus. Phenotypic traits also include characteristics such as longevity, appearance (e.g., baldness, obesity), strength, speed, endurance, fertility, and susceptibility or receptivity to particular drugs or therapeutic treatments.

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Such correlations can be exploited in several ways. In the case of a strong correlation between a polymorphic form and a disease for which treatment is available, detection of the polymorphic form set in a human or animal patient may justify immediate administration of treatment, or at least the institution of regular monitoring of the patient. Detection of a polymorphic form correlated with serious disease in a couple contemplating a family may also be valuable to the couple in their reproductive decisions. For example, the female partner might elect to undergo in vitro fertilization to avoid the possibility of transmitting such a polymorphism from her husband to her offspring. In the case of a weaker, but still statistically significant correlation between a polymorphic set and human disease, immediate therapeutic intervention or monitoring may not be justified. Nevertheless, the patient can be motivated to begin simple life-style changes (e.g., diet, exercise) that can be accomplished at little cost to the patient but confer potential benefits in reducing the risk of conditions to which the patient may have increased susceptibility by virtue of variant alleles. After determining polymorphic form(s) present in an individual at one or more polymorphic sites, this information can be used in a number of methods.

Determination of which polymorphic forms occupy a set of polymorphic sites in an individual identifies a set of polymorphic forms that distinguishes the individual. See generally National Research Council, *The Evaluation of Forensic DNA Evidence* (Eds. Pollard et al., National Academy Press, DC, 1996). Since the polymorphic sites are within a 50,000 bp region in the human genome, the probability of recombination between these

polymorphic sites is low. That low probability means the haplotype (the set of all 10 polymorphic sites) set forth in this application should be inherited without change for at least several generations. The more sites that are analyzed the lower the probability that the set of polymorphic forms in one individual is the same as that in an unrelated individual.

Preferably, if multiple sites are analyzed, the sites are unlinked. Thus, polymorphisms of the invention are often used in conjunction with polymorphisms in distal genes. Preferred polymorphisms for use in forensics are diallelic because the population frequencies of two polymorphic forms can usually be determined with greater accuracy than those of multiple polymorphic forms at multi-allelic loci.

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The capacity to identify a distinguishing or unique set of forensic markers in an individual is useful for forensic analysis. For example, one can determine whether a blood sample from a suspect matches a blood or other tissue sample from a crime scene by determining whether the set of polymorphic forms occupying selected polymorphic sites is the same in the suspect and the sample. If the set of polymorphic markers does not match between a suspect and a sample, it can be concluded (barring experimental error) that the suspect was not the source of the sample. If the set of markers does match, one can conclude that the DNA from the suspect is consistent with that found at the crime scene. If frequencies of the polymorphic forms at the loci tested have been determined (e.g., by analysis of a suitable population of individuals), one can perform a statistical analysis to determine the probability that a match of suspect and crime scene sample would occur by chance.

p(ID) is the probability that two random individuals have the same polymorphic or allelic form at a given polymorphic site. In diallelic loci, four genotypes are possible: AA, AB, BA, and BB. If alleles A and B occur in a haploid genome of the organism with frequencies x and y, the probability of each genotype in a diploid organism are (see WO 95/12607):

Homozygote: $p(AA)=x^2$

Homozygote: $p(BB)=y^2=(1-x)^2$

Single Heterozygote: p(AB)=p(BA)=xy=x(1-x)

Both Heterozygotes: p(AB+BA)=2xy=2x(1-x)

The probability of identity at one locus (i.e, the probability that two individuals, picked at random from a population will have identical polymorphic forms at a given locus) is given by the equation:

$$p(ID)=(x^2)^{2+}(2xy)^{2+}(y^2)^2$$
.

These calculations can be extended for any number of polymorphic forms at a given locus. For example, the probability of identity p(ID) for a 3-allele system where the alleles have the frequencies in the population of x, y and z, respectively, is equal to the sum of the squares of the genotype frequencies:

$$p(ID)=x^{4+}(2xy)^{2+}(2yz)^{2+}(2xz)^{2+}z^{4+}y^{4}$$

In a locus of n alleles, the appropriate binomial expansion is used to calculate p(ID) and p(exc).

The cumulative probability of identity (cum p(ID)) for each of multiple unlinked loci is determined by multiplying the probabilities provided by each locus:

$$cum p(ID)=p(ID1)p(ID2)p(ID3) \dots p(IDn)$$

The cumulative probability of non-identity for n loci (i.e. the probability that two random individuals will be different at 1 or more loci) is given by the equation:

$$cum p(nonID)=1-cum p(ID).$$

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If several polymorphic loci are tested, the cumulative probability of non-identity for random individuals becomes very high (e.g., one billion to one). Such probabilities can be taken into account together with other evidence in determining the guilt or innocence of the suspect.

The object of paternity testing is usually to determine whether a male is the father of a child. In most cases, the mother of the child is known and thus, the mother's contribution to the child's genotype can be traced. Paternity testing investigates whether the part of the child's genotype not attributable to the mother is consistent with that of the putative father. Paternity testing can be performed by analyzing sets of polymorphisms in the putative father and the child.

If the set of polymorphisms in the child attributable to the father does not match the putative father, it can be concluded, barring experimental error, that the putative father is not the real father. If the set of polymorphisms in the child attributable to the father does match the set of polymorphisms of the putative father, a statistical calculation can be performed to determine the probability of coincidental match.

The probability of parentage exclusion (representing the probability that a random male will have a polymorphic form at a given polymorphic site that makes him incompatible as the father) is given by the equation (see WO 95/12607):

$$p(exc)=xy(1-xy)$$

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where x and y are the population frequencies of alleles A and B of a diallelic polymorphic site. (At a triallelic site p(exc)=xy(1-xy)+ yz(1-yz)+ xz(1-xz)+ 3xyz(1-xyz))), where x, y and z and the respective population frequencies of alleles A, B and C). The probability of non-exclusion is:

$$p(non-exc)=1-p(exc)$$

15 The cumulative probability of non-exclusion (representing the value obtained when n loci are used) is thus:

$$cum p(non-exc)=p(non-exc1)p(non-exc2)p(non-exc3)...p(non-excn)$$

The cumulative probability of exclusion for n loci (representing the probability that a random male will be excluded) is:

20 cum p(exc)=1-cum p(non-exc).

If several polymorphic loci are included in the analysis, the cumulative probability of exclusion of a random male is very high. This probability can be taken into account in assessing the liability of a putative father whose polymorphic marker set matches the child's polymorphic marker set attributable to his/her father.

The polymorphisms of the invention may contribute to the phenotype of an organism in different ways. Some polymorphisms occur within a protein coding sequence and contribute to phenotype by affecting protein structure. The effect may be neutral, beneficial or detrimental, or both beneficial and detrimental, depending on the circumstances. For

example, a heterozygous sickle cell mutation confers resistance to malaria, but a homozygous sickle cell mutation is usually lethal. Other polymorphisms occur in noncoding regions but may exert phenotypic effects indirectly via influence on replication, transcription, and translation. A single polymorphism may affect more than one phenotypic trait. Likewise, a single phenotypic trait may be affected by polymorphisms in different genes. Further, some polymorphisms predispose an individual to a distinct mutation that is causally related to a certain phenotype.

Phenotypic traits include diseases that have known but hitherto unmapped genetic components. Phenotypic traits also include symptoms of, or susceptibility to, multifactorial diseases of which a component is or may be genetic, such as autoimmune diseases, inflammation, cancer, diseases of the nervous system, and infection by pathogenic microorganisms. Some examples of autoimmune diseases include rheumatoid arthritis, multiple sclerosis, diabetes (insulin-dependent and non-independent), systemic lupus erythematosus and Graves disease. Some examples of cancers include cancers of the bladder, brain, breast, colon, esophagus, kidney, leukemia, liver, lung, oral cavity, ovary, pancreas, prostate, skin, stomach and uterus. Phenotypic traits also include characteristics such as longevity, appearance (e.g., baldness, obesity), strength, speed, endurance, fertility, and susceptibility or receptivity to particular drugs or therapeutic treatments.

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Correlation is performed for a population of individuals who have been tested for the presence or absence of a phenotypic trait of interest and for polymorphic markers sets. To perform such analysis, the presence or absence of a set of polymorphisms (i.e. a polymorphic set) is determined for a set of the individuals, some of whom exhibit a particular trait, and some of which exhibit lack of the trait. The alleles of each polymorphism of the set are then reviewed to determine whether the presence or absence of a particular allele is associated with the trait of interest. Correlation can be performed by standard statistical methods such as a -squared test and statistically significant correlations between polymorphic form(s) and phenotypic characteristics are noted. For example, it might be found that the presence of allele A1 at polymorphism A correlates with heart disease. As a further example, it might be found that the combined presence of allele A1 at polymorphism A and allele B1 at polymorphism B correlates with increased milk production of a farm animal.

Such correlations can be exploited in several ways. In the case of a strong correlation between a set of one or more polymorphic forms and a disease for which treatment is

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available, detection of the polymorphic form set in a human or animal patient may justify immediate administration of treatment, or at least the institution of regular monitoring of the patient. Detection of a polymorphic form correlated with serious disease in a couple contemplating a family may also be valuable to the couple in their reproductive decisions. For example, the female partner might elect to undergo in vitro fertilization to avoid the possibility of transmitting such a polymorphism from her husband to her offspring. In the case of a weaker, but still statistically significant correlation between a polymorphic set and human disease, immediate therapeutic intervention or monitoring may not be justified. Nevertheless, the patient can be motivated to begin simple life-style changes (e.g., diet, exercise) that can be accomplished at little cost to the patient but confer potential benefits in reducing the risk of conditions to which the patient may have increased susceptibility by virtue of variant alleles. Identification of a polymorphic set in a patient correlated with enhanced receptiveness to one of several treatment regimes for a disease indicates that this treatment regime should be followed.

For animals and plants, correlations between characteristics and phenotype are useful for breeding for desired characteristics. For example, Beitz et al., U.S. Pat. No. 5,292,639 discuss use of bovine mitochondrial polymorphisms in a breeding program to improve milk production in cows. To evaluate the effect of mtDNA D-loop sequence polymorphism on milk production, each cow was assigned a value of 1 if variant or 0 if wild type with respect to a prototypical mitochondrial DNA sequence at each of 17 locations considered.

The previous section concerns identifying correlations between phenotypic traits and polymorphisms that directly or indirectly contribute to those traits. The present section describes identification of a physical linkage between a genetic locus associated with a trait of interest and polymorphic markers that are not associated with the trait, but are in physical proximity with the genetic locus responsible for the trait and co-segregate with it. Such analysis is useful for mapping a genetic locus associated with a phenotypic trait to a chromosomal position, and thereby cloning gene(s) responsible for the trait. See Lander et al., *Proc. Natl. Acad. Sci.* (USA) 83, 7353-7357 (1986); Lander et al., *Proc. Natl. Acad. Sci.* (USA) 84, 2363-2367 (1987); Donis-Keller et al., *Cell* 51, 319-337 (1987); Lander et al., *Genetics* 121, 185-199 (1989)). Genes localized by linkage can be cloned by a process known as directional cloning. See Wainwright, *Med. J. Australia* 159, 170-174 (1993); Collins, *Nature Genetics* 1, 3-6 (1992) (each of which is incorporated by reference in its entirety for all purposes).

Linkage studies are typically performed on members of a family. Available members of the family are characterized for the presence or absence of a phenotypic trait and for a set of polymorphic markers. The distribution of polymorphic markers in an informative meiosis is then analyzed to determine which polymorphic markers co-segregate with a phenotypic trait. See, e.g., Kerem et al., *Science* 245, 1073-1080 (1989); Monaco et al., *Nature* 316, 842 (1985); Yamoka et al., *Neurology* 40, 222-226 (1990); Rossiter et al., *FASEB Journal* 5, 21-27 (1991).

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Linkage is analyzed by calculation of LOD (log of the odds) values. A lod value is the relative likelihood of obtaining observed segregation data for a marker and a genetic locus when the two are located at a recombination fraction , versus the situation in which the two are not linked, and thus segregating independently (Thompson & Thompson, Genetics in Medicine (5th ed, W.B. Saunders Company, Philadelphia, 1991); Strachan, "Mapping the human genome" in The Human Genome (BIOS Scientific Publishers Ltd, Oxford), Chapter 4). A series of likelihood ratios are calculated at various recombination fractions (), ranging from =0.0 (coincident loci) to =0.50 (unlinked). Thus, the likelihood at a given value of is: probability of data if loci linked at to probability of data if loci unlinked. The computed likelihood is usually expressed as the log10 of this ratio (i.e., a lod score). For example, a lod score of 3 indicates 1000:1 odds against an apparent observed linkage being a coincidence. The use of logarithms allows data collected from different families to be combined by simple addition. Computer programs are available for the calculation of lod scores for differing values of (e.g., LIPED, MLINK (Lathrop.Proc. Nat. Acad. Sci. (USA) 81, 3443-3446 (1984)). For any particular lod score, a recombination fraction may be determined from mathematical tables. See Smith et al., Mathematical tables for research workers in human genetics (Churchill, London, 1961); Smith, Ann. Hum. Genet. 32, 127-150 (1968). The value of at which the lod score is the highest is considered to be the best estimate of the recombination fraction.

Positive lod score values suggest that the two loci are linked, whereas negative values suggest that linkage is less likely (at that value of) than the possibility that the two loci are unlinked. By convention, a combined lod score of + 3 or greater (equivalent to greater than 1000:1 odds in favor of linkage) is considered definitive evidence that two loci are linked. Similarly, by convention, a negative lod score of -2 or less is taken as definitive evidence against linkage of the two loci being compared. Negative linkage data are useful in

excluding a chromosome or a segment thereof from consideration. The search focuses on the remaining non-excluded chromosomal locations.

The invention further provides transgenic nonhuman animals capable of expressing an exogenous variant gene and/or having one or both alleles of an endogenous variant gene inactivated. Expression of an exogenous variant gene is usually achieved by operably linking the gene to a promoter and optionally an enhancer, and microinjecting the construct into a zygote. See Hogan et al., "Manipulating the Mouse Embryo, A Laboratory Manual," Cold Spring Harbor Laboratory. (1989). Inactivation of endogenous variant genes can be achieved by forming a transgene in which a cloned variant gene is inactivated by insertion of a positive selection marker. See Capecchi, Science 244, 1288-1292 The transgene is then introduced into an embryonic stem cell, where it undergoes homologous recombination with an endogenous variant gene. Mice and other rodents are preferred animals. Such animals provide useful drug screening systems.

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The invention further provides methods for assessing the pharmacogenomic susceptibility of a subject harboring a single nucleotide polymorphism to a particular pharmaceutical compound, or to a class of such compounds. Genetic polymorphism in drugmetabolizing enzymes, drug transporters, receptors for pharmaceutical agents, and other drug targets have been correlated with individual differences based on distinction in the efficacy and toxicity of the pharmaceutical agent administered to a subject. Pharmocogenomic characterization of a subjects susceptibility to a drug enhances the ability to tailor a dosing regimen to the particular genetic constitution of the subject, thereby enhancing and optimizing the therapeutic effectiveness of the therapy.

In cases in which a cSNP leads to a polymorphic protein that is ascribed to be the cause of a pathological condition, method of treating such a condition includes administering to a subject experiencing the pathology the wild type cognate of the polymorphic protein.

Once administered in an effective dosing regimen, the wild type cognate provides complementation or remediation of the defect due to the polymorphic protein. The subject's condition is ameliorated by this protein therapy.

A subject suspected of suffering from a pathology ascribable to a polymorphic protein that arises from a cSNP is to be diagnosed using any of a variety of diagnostic methods capable of identifying the presence of the cSNP in the nucleic acid, or of the cognate

polymorphic protein, in a suitable clinical sample taken from the subject. Once the presence of the cSNP has been ascertained, and the pathology is correctable by administering a normal or wild-type gene, the subject is treated with a pharmaceutical composition that includes a nucleic acid that harbors the correcting wild-type gene, or a fragment containing a correcting sequence of the wild-type gene. Non-limiting examples of ways in which such a nucleic acid may be administered include incorporating the wild-type gene in a viral vector, such as an adenovirus or adeno associated virus, and administration of a naked DNA in a pharmaceutical composition that promotes intracellular uptake of the administered nucleic acid. Once the nucleic acid that includes the gene coding for the wild-type allele of the polymorphism is incorporated within a cell of the subject, it will initiate *de novo* biosynthesis of the wild-type gene product. If the nucleic acid is further incorporated into the genome of the subject, the treatment will have long-term effects, providing *de novo* synthesis of the wild-type protein for a prolonged duration. The synthesis of the wild-type protein in the cells of the subject will contribute to a therapeutic enhancement of the clinical condition of the subject.

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A subject suffering from a pathology ascribed to a SNP may be treated so as to correct the genetic defect. (See Kren et al., Proc. Natl. Acad. Sci. USA 96:10349-10354 (1999)). Such a subject is identified by any method that can detect the polymorphism in a sample drawn from the subject. Such a genetic defect may be permanently corrected by administering to such a subject a nucleic acid fragment incorporating a repair sequence that supplies the wild-type nucleotide at the position of the SNP. This site-specific repair sequence encompasses an RNA/DNA oligonucleotide which operates to promote endogenous repair of a subject's genomic DNA. Upon administration in an appropriate vehicle, such as a complex with polyethylenimine or encapsulated in anionic liposomes, a genetic defect leading to an inborn pathology may be overcome, as the chimeric oligonucleotides induces incorporation of the wild-type sequence into the subject's genome. Upon incorporation, the wild-type gene product is expressed, and the replacement is propagated, thereby engendering a permanent repair.

The invention further provides kits comprising at least one allele-specific oligonucleotide as described above. Often, the kits contain one or more pairs of allele-specific oligonucleotides hybridizing to different forms of a polymorphism. In some kits, the allele-specific oligonucleotides are provided immobilized to a substrate. For example, the same substrate can comprise allele-specific oligonucleotide probes for detecting at least 10, 100, 1000 or all of the polymorphisms shown in the Table. Optional additional

components of the kit include, for example, restriction enzymes, reverse-transcriptase or polymerase, the substrate nucleoside triphosphates, means used to label (for example, an avidin-enzyme conjugate and enzyme substrate and chromogen if the label is biotin), and the appropriate buffers for reverse transcription, PCR, or hybridization reactions. Usually, the kit also contains instructions for carrying out the hybridizing methods.

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Several aspects of the present invention rely on having available the polymorphic proteins encoded by the nucleic acids comprising a SNP of the inventions. There are various methods of isolating these nucleic acid sequences. For example, DNA is isolated from a genomic or cDNA library using labeled oligonucleotide probes having sequences complementary to the sequences disclosed herein.

Such probes can be used directly in hybridization assays. Alternatively probes can be designed for use in amplification techniques such as PCR.

To prepare a cDNA library, mRNA is isolated from tissue such as heart or pancreas, preferably a tissue wherein expression of the gene or gene family is likely to occur. cDNA is prepared from the mRNA and ligated into a recombinant vector. The vector is transfected into a recombinant host for propagation, screening and cloning. Methods for making and screening cDNA libraries are well known, See Gubler, U. and Hoffman, B.J. Gene 25:263-269 (1983) and Sambrook et al.

For a genomic library, for example, the DNA is extracted from tissue and either mechanically sheared or enzymatically digested to yield fragments of about 12-20 kb. The fragments are then separated by gradient centrifugation from undesired sizes and are constructed in bacteriophage lambda vectors. These vectors and phage are packaged *in vitro*, as described in Sambrook, et al. Recombinant phage are analyzed by plaque hybridization as described in Benton and Davis, Science 1'96:180-182 (1977). Colony hybridization is carried out as generally described in M. Grunstein et al. Proc. Natl. Acad. Sci. USA. 72:3961-3965 (1975). DNA of interest is identified in either cDNA or genomic libraries by its ability to hybridize with nucleic acid probes, for example on Southern blots, and these DNA regions are isolated by standard methods familiar to those of skill in the art. See Sambrook, et al.

In PCR techniques, oligonucleotide primers complementary to the two 3' borders of the DNA region to be amplified are synthesized. The polymerase chain reaction is then carried out using the two primers. See PCR Protocols: a Guide to Methods and Applications

(Innis, M, Gelfand, D., Sninsky, J. and White, T., eds.), Academic Press, San Diego (1990). Primers can be selected to amplify the entire regions encoding a full-length sequence of interest or to amplify smaller DNA. segments as desired. PCR can be used in a variety of protocols to isolate cDNA's encoding a sequence of interest. In these protocols, appropriate primers and probes for amplifying DNA encoding a sequence of interest are generated from analysis of the DNA sequences listed herein. Once such regions are PCR-amplified, they can be sequenced and oligonucleotide probes can be prepared from the sequence.

Once DNA encoding a sequence comprising a cSNP is isolated and cloned, one can express the encoded polymorphic proteins in a variety of recombinantly engineered cells. It is expected that those of skill in the art are knowledgeable in the numerous expression systems available for expression of DNA encoding a sequence of interest. No attempt to describe in detail the various methods known for the expression of proteins in prokaryotes or eukaryotes is made here.

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In brief summary, the expression of natural or synthetic nucleic acids encoding a sequence of interest will typically be achieved by operably linking the DNA or cDNA to a promoter (which is either constitutive or inducible), followed by incorporation into an expression vector. The vectors can be suitable for replication and integration in either prokaryotes or eukaryotes. Typical expression vectors contain, initiation sequences, transcription and translation terminators, and promoters useful for regulation of the expression of a polynucleotide sequence of interest. To obtain high level expression of a cloned gene, it is desirable to construct expression plasmids which contain, at the minimum, a strong promoter to direct transcription, a ribosome binding site for translational initiation, and a transcription/translation terminator. The expression vectors may also comprise generic expression cassettes containing at least one independent terminator sequence, sequences permitting replication of the plasmid in both eukaryotes and prokaryotes, i.e., shuttle vectors, and selection markers for both prokaryotic and eukaryotic systems. See Sambrook et al.

A variety of prokaryotic expression systems may be used to express the polymorphic proteins of the invention. Examples include *E. coli*, Bacillus, Streptomyces, and the like.

It is preferred to construct expression plasmids which contain, at the minimum, a strong promoter to direct transcription, a ribosome binding site for translational initiation, and a transcription/translation terminator. Examples of regulatory regions suitable for this

purpose in *E. coli* are the promoter and operator region of the *E. coli* tryptophan biosynthetic pathway as described by Yanofsky, C., J. Bacterial. 158:1018-1024 (1984) and the leftward promoter of phage lambda (P) as described by Λ, I. and Hagen, D., Ann. Rev. Genet. 14:399-445 (1980). The inclusion of selection markers in DNA vectors transformed in *E. coli* is also useful. Examples of such markers include genes specifying resistance to ampicillin, tetracycline, or chloramphenicol. See Sambrook et al. for details concerning selection markers for use in *E. coli*.

To enhance proper folding of the expressed recombinant protein, during purification from *E. coli*, the expressed protein may first be denatured and then renatured. This can be accomplished by solubilizing the bacterially produced proteins in a chaotropic agent such as guanidine HCI and reducing all the cysteine residues with a reducing agent such as beta-mercaptoethanol. The protein is then renatured, either by slow dialysis or by gel filtration. See U.S. Patent No. 4,511,503. Detection of the expressed antigen is achieved by methods known in the art as radioimmunoassay, or Western blotting techniques or immunoprecipitation. Purification from *E. coli* can be achieved following procedures such as those described in U.S. Patent No. 4,511,503.

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Any of a variety of eukaryotic expression systems such as yeast, insect cell lines, bird, fish, and mammalian cells, may also be used to express a polymorphic protein of the invention. As explained briefly below, a nucleotide sequence harboring a cSNP may be expressed in these eukaryotic systems. Synthesis of heterologous proteins in yeast is well known. Methods in Yeast Genetics, Sherman, F., et al., Cold Spring Harbor Laboratory, (1982) is a well recognized work describing the various methods available to produce the protein in yeast. Suitable vectors usually have expression control sequences, such as promoters, including 3-phosphogtycerate kinase or other glycolytic enzymes, and an origin of replication, termination sequences and the like as desired. For instance, suitable vectors are described in the literature (Botstein, et al., Gene 8:17-24 (1979); Broach, et al., Gene 8:121-133 (1979)).

Two procedures are used in transforming yeast cells. In one case, yeast cells are first converted into protoplasts using zymolyase, lyticase or glusulase, followed by addition of DNA and polyethylene glycol (PEG). The PEG-treated protoplasts are then regenerated in a 3% agar medium under selective conditions. Details of this procedure are given in the papers by J.D. Beggs, Nature (London) 275:104-109 (1978); and Hinnen, A., et al., Proc. Natl.

Acad. Sci. USA, 75:1929-1933 (1978). The second procedure does not involve removal of the cell wall. Instead the cells are treated with lithium chloride or acetate and PEG and put on selective plates (Ito, H., et al., J. Bact, 153163-168 (1983)). cells and applying standard protein isolation techniques to the lysates:.

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The purification process can be monitored by using Western blot techniques or radioimmunoassay or other standard techniques. The sequences encoding the proteins of the invention can also be ligated to various immunoassay expression vectors for use in transforming cell cultures of, for instance, mammalian, insect, bird or fish origin. Illustrative of cell cultures useful for the production of the polypeptides are mammalian cells.

Mammalian cell systems often will be in the form of monolayers of cells although mammalian cell suspensions may also be used. A number of suitable host cell lines capable of expressing intact proteins have been developed in the art, and include the HEK293, BHK21, and CHO cell lines, and various human cells such as COS cell lines, HeLa cells, myeloma cell lines, Jurkat cells, etc. Expression vectors for these cells can include expression control sequences, such as an origin of replication, a promoter (e.g., the CMV promoter, a HSV tk promoter or pgk (phosphoglycerate kinase) promoter), an enhancer (Queen et al. Immunol. Rev. 89:49 (1986)) and necessary processing information sites, such as ribosome binding sites, RNA splice sites, polyadenylation sites (e.g., an SV40 large T Ag poly A addition site), and transcriptional terminator sequences.

Other animal cells are available, for instance, from the American Type Culture Collection Catalogue of Cell Lines and Hybridomas (7th edition, (1992)). Appropriate vectors for expressing the proteins of the invention in insect cells are usually derived from baculovirus. Insect cell lines include mosquito larvae, silkworm, armyworm, moth and Drosophila cell lines such as a Schneider cell line (See Schneider J. Embryol. Exp. Morphol., 27:353-365 (1987). As indicated above, the vector, e.g., a plasmid, which is used to transform the host cell, preferably contains DNA sequences to initiate transcription and sequences to control the translation of the protein. These sequences are referred to as expression control sequences. As with yeast, when higher animal host cells are employed, polyadenylation or transcription terminator sequences from known mammalian genes need to be incorporated into the vector. An example of a terminator sequence is the polyadenylation sequence from the bovine growth hormone gene. Sequences for accurate splicing of the transcript may also be included. An example of a splicing sequence is the VP1 intron from SV4O (Sprague, J. et a/., J. Virol. 45: 773-781 (1983)). Additionally, gene sequences to

control replication in the host cell may be Saveria-Campo, M., 1985, "Bovine Papilloma virus DNA a Eukaryotic Cloning Vector" in DNA Cloning Vol. II a Practical Approach Ed. D.M. Glover, IRL Press, Arlington, Virginia pp. 213-238. The host cells are competent or rendered competent for transformation by various means. There are several well-known methods of introducing DNA into animal cells. These include: calcium phosphate precipitation, fusion of the recipient cells with bacterial protoplasts containing the DNA, treatment of the recipient cells with liposomes containing the DNA, DEAE dextran, electroporation and micro-injection of the DNA directly into the cells.

The transformed cells are cultured by means well known in the art (Biochemical Methods in Cell Culture and Virology, Kuchler, R.J., Dowden, Hutchinson and Ross, Inc., (1977)). The expressed polypeptides are isolated from cells grown as suspensions or as monolayers. The latter are recovered by well known mechanical, chemical or enzymatic means.

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General methods of expressing recombinant proteins are also known and are exemplified in R. Kaufman, Methods in Enzymology 185, 537-566 (1990). As defined herein "operably linked" refers to linkage of a promoter upstream from a DNA sequence such that the promoter mediates transcription of the DNA sequence. Specifically, "operably linked" means that the isolated polynucleotide of the invention and an expression control sequence are situated within a vector or cell in such a way that the gene encoding the protein is expressed by a host cell which has been transformed (transfected) with the ligated polynucleotide/expression sequence. The term "vector", refers to viral expression systems, autonomous self-replicating circular DNA (plasmids), and includes both expression and nonexpression plasmids.

The term "gene" as used herein is intended to refer to a nucleic acid sequence which encodes a polypeptide. This definition includes various sequence polymorphisms, mutations, and/or sequence variants wherein such alterations do not affect the function of the gene product. The term "gene" is intended to include not only coding sequences but also regulatory regions such as promoters, enhancers, termination regions and similar untranslated nucleotide sequences. The term further includes all introns and other DNA sequences spliced from the mRNA transcript, along with variants resulting from alternative splice sites.

A number of types of cells may act as suitable host cells for expression of the protein. Mammalian host cells include, for example, monkey COS cells, Chinese Hamster Ovary (CHO) cells, human kidney 293 cells, human epidermal A43 1 cells, human Co10205 cells, 3T3 cells, CV-1 cells, other transformed primate cell lines, normal diploid cells, cell strains derived from in vitro culture of primary tissue, primary explants, HeLa cells, mouse L cells, BHK, HL-60, U937, HaK or Jurkat cells. Alternatively, it may be possible to produce the protein in lower eukaryotes such as yeast or in prokaryotes such as bacteria. Potentially suitable yeast strains include Saccharomyces cerevisiae, Schizosaccharomyces pombe, Kluyveromyces strains, Candida or any yeast strain capable of expressing heterologous proteins. Potentially suitable bacterial strains include Escherichia coli, Bacillus subtilis, Salmonella typhimurium, or any bacterial strain capable of expressing heterologous proteins. If the protein is made in yeast or bacteria, it may be necessary to modify the protein produced therein, for example by phosphorylation or glycosylation of the appropriate sites, in order to obtain the functional protein.

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The protein may also be produced by operably linking the isolated polynucleotide of the invention to suitable control sequences in one or more insect expression vectors, and employing an insect expression system. Materials and methods for baculovirus/insect cell expression systems are commercially available in kit form from, e.g., Invitrogen, San Diego, California, U.S.A. (the MaxBac© kit), and such methods are well known in the art, as described in Summers and Smith, Texas Agricultural Experiment Station Bulletin No. 1555 (1987), incorporated herein by reference. As used herein, an insect cell capable of expressing a polynucleotide of the present invention is "transformed." The protein of the invention may be prepared by culturing transformed host cells under culture conditions suitable to express the recombinant protein.

The polymorphic protein of the invention may also be expressed as a product of transgenic animals, e.g., as a component of the milk of transgenic cows, goats, pigs, or sheep which are characterized by somatic or germ cells containing a nucleotide sequence encoding the protein. The protein may also be produced by known conventional chemical synthesis. Methods for constructing the proteins of the present invention by synthetic means are known to those skilled in the art.

The polymorphic proteins produced by recombinant DNA technology may be purified by techniques commonly employed to isolate or purify recombinant proteins. Recombinantly

produced proteins can be directly expressed or expressed as a fusion protein. The protein is then purified by a combination of cell lysis (e.g., sonication) and affinity chromatography. For fusion products, subsequent digestion of the fusion protein with an appropriate proteolytic enzyme releases the desired polypeptide. The polypeptides of this invention may be purified to substantial purity by standard techniques well known in the art, including selective precipitation with such substances as ammonium sulfate, column chromatography, immunopurification methods, and others. See, for instance, R. Scopes, Protein Purification: Principles and Practice, Springer-Verlag: New York (1982), incorporated herein by reference. For example, in an embodiment, antibodies may be raised to the proteins of the invention as described herein. Cell membranes are isolated from a cell line expressing the recombinant protein, the protein is extracted from the membranes and immunoprecipitated. The proteins may then be further purified by standard protein chemistry techniques as described above.

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The resulting expressed protein may then be purified from such culture (i.e., from culture medium or cell extracts) using known purification processes, such as gel filtration and ion exchange chromatography. The purification of the protein may also include an affinity column containing agents which will bind to the protein; one or more column steps over such affinity resins as concanavalin A-agarose, heparin-Toyopearl@ or Cibacrom blue 3GA Sepharose B; one or more steps involving hydrophobic interaction chromatography using such resins as phenyl ether, butyl ether, or propyl ether; or immunoaffinity chromatography. Alternatively, the protein of the invention may also be expressed in a form which will facilitate purification. For example, it may be expressed as a fusion protein, such as those of maltose binding protein (MBP), glutathione-S-transferase (GST) or thioredoxin (TRX). Kits for expression and purification of such fusion proteins are commercially available from New England BioLab (Beverly, MA), Pharmacia (Piscataway, NJ) and InVitrogen, respectively. The protein can also be tagged with an epitope and subsequently purified by using a specific antibody directed to such epitope. One such epitope ("Flag") is commercially available from Kodak (New Haven, CT). Finally, one or more reverse-phase high performance liquid chromatography (RP- HPLC) steps employing hydrophobic RP-HPLC media, e.g., silica gel having pendant methyl or other aliphatic groups, can be employed to further purify the protein. Some or all of the foregoing purification steps, in various combinations, can also be employed to provide a substantially homogeneous isolated recombinant protein. The protein thus purified is substantially free of other

mammalian proteins and is defined in accordance with the present invention as an "isolated protein."

The term "antibody" as used herein refers to immunoglobulin molecules and immunologically active portions of immunoglobulin molecules, *i.e.*, molecules that contain an antigen binding site that specifically binds (immunoreacts with) an antigen, such as polymorphic. Such antibodies include, but are not limited to, polyclonal, monoclonal, chimeric, single chain, F_{ab} and $F_{(ab)2}$ fragments, and an F_{ab} expression library. In a specific embodiment, antibodies to human polymorphic proteins are disclosed.

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The phrase "specifically binds to", "immunospecifically binds to" or is "specifically immunoreactive with", an antibody when referring to a protein or peptide, refers to a binding reaction which is determinative of the presence of the protein in the presence of a heterogeneous population of proteins and other biological materials. Thus, for example, under designated immunoassay conditions, the specified antibodies bind to a particular protein and do not bind in a significant amount to other proteins present in the sample. Specific binding to an antibody under such conditions may require an antibody that is selected for its specificity for a particular protein. Of particular interest in the present invention is an antibody that binds immunospecifically to a polymorphic protein but not to its cognate wild type allelic protein, or vice versa. A variety of immunoassay formats may be used to select antibodies specifically immunoreactive with a particular protein. For example, solid-phase ELISA immunoassays are routinely used to select monoclonal antibodies specifically immunoreactive with a protein. See Harlow and Lane (1988) Antibodies, a Laboratory Manual, Cold Spring Harbor Publications, New York, for a description of immunoassay formats and conditions that can be used to determine specific immunoreactivity.

Polyclonal and/or monoclonal antibodies that immunospecifically bind to polymorphic gene products but not to the corresponding prototypical or "wild-type" gene products are also provided. Antibodies can be made by injecting mice or other animals with the variant gene product or synthetic peptide. Monoclonal antibodies are screened as are described, for example, in Harlow & Lane, Antibodies, A Laboratory Manual, Cold Spring Harbor Press, New York (1988); Goding, Monoclonal antibodies, Principles and Practice (2d ed.) Academic Press, New York (1986). Monoclonal antibodies are tested for specific

immunoreactivity with a variant gene product and lack of immunoreactivity to the corresponding prototypical gene product.

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An isolated polymorphic protein, or a portion or fragment thereof, can be used as an immunogen to generate the antibody that bind the polymorphic protein using standard techniques for polyclonal and monoclonal antibody preparation. The full-length polymorphic protein can be used or, alternatively, the invention provides antigenic peptide fragments of polymorphic for use as immunogens. The antigenic peptide of a polymorphic protein of the invention comprises at least 8 amino acid residues of the amino acid sequence encompassing the polymorphic amino acid and encompasses an epitope of the polymorphic protein such that an antibody raised against the peptide forms a specific immune complex with the polymorphic protein. Preferably, the antigenic peptide comprises at least 10 amino acid residues, more preferably at least 15 amino acid residues, even more preferably at least 20 amino acid residues, and most preferably at least 30 amino acid residues. Preferred epitopes encompassed by the antigenic peptide are regions of polymorphic that are located on the surface of the protein, *e.g.*, hydrophilic regions.

For the production of polyclonal antibodies, various suitable host animals (e.g., rabbit, goat, mouse or other mammal) may be immunized by injection with the polymorphic protein. An appropriate immunogenic preparation can contain, for example, recombinantly expressed polymorphic protein or a chemically synthesized polymorphic polypeptide. The preparation can further include an adjuvant. Various adjuvants used to increase the immunological response include, but are not limited to, Freund's (complete and incomplete), mineral gels (e.g., aluminum hydroxide), surface active substances (e.g., lysolecithin, pluronic polyols, polyanions, peptides, oil emulsions, dinitrophenol, etc.), human adjuvants such as Bacille Calmette-Guerin and Corynebacterium parvum, or similar immunostimulatory agents. If desired, the antibody molecules directed against polymorphic proteins can be isolated from the mammal (e.g., from the blood) and further purified by well known techniques, such as protein A chromatography, to obtain the IgG fraction.

The term "monoclonal antibody" or "monoclonal antibody composition", as used herein, refers to a population of antibody molecules that originates from the clone of a singly hybridoma cell, and that contains only one type of antigen binding site capable of immunoreacting with a particular epitope of a polymorphic protein. A monoclonal antibody composition thus typically displays a single binding affinity for a particular polymorphic

protein with which it immunoreacts. For preparation of monoclonal antibodies directed towards a particular polymorphic protein, or derivatives, fragments, analogs or homologs thereof, any technique that provides for the production of antibody molecules by continuous cell line culture may be utilized. Such techniques include, but are not limited to, the hybridoma technique (see Kohler & Milstein, 1975 Nature 256: 495-497); the trioma technique; the human B-cell hybridoma technique (see Kozbor, et al., 1983 Immunol Today 4: 72) and the EBV hybridoma technique to produce human monoclonal antibodies (see Cole, et al., 1985 In: Monoclonal Antibodies and Cancer Therapy, Alan R. Liss, Inc., pp. 77-96). Human monoclonal antibodies may be utilized in the practice of the present invention and may be produced by using human hybridomas (see Cote, et al., 1983. Proc Natl Acad Sci USA 80: 2026-2030) or by transforming human B-cells with Epstein Barr Virus in vitro (see Cole, et al., 1985 In: Monoclonal Antibodies and Cancer Therapy, Alan R. Liss, Inc., pp. 77-96).

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According to the invention, techniques can be adapted for the production of single-chain antibodies specific to a polymorphic protein (see e.g., U.S. Patent No. 4,946,778). In addition, methodologies can be adapted for the construction of F_{ab} expression libraries (see e.g., Huse, et al., 1989 Science 246: 1275-1281) to allow rapid and effective identification of monoclonal F_{ab} fragments with the desired specificity for a polymorphic protein or derivatives, fragments, analogs or homologs thereof. Non-human antibodies can be "humanized" by techniques well known in the art. See e.g., U.S. Patent No. 5,225,539. Antibody fragments that contain the idiotypes to a polymorphic protein may be produced by techniques known in the art including, but not limited to: (i) an $F_{(ab)}$ fragment produced by pepsin digestion of an antibody molecule; (ii) an F_{ab} fragment generated by reducing the disulfide bridges of an $F_{(ab)}$ fragment; (iii) an F_{ab} fragment generated by the treatment of the antibody molecule with papain and a reducing agent and (iv) F_{v} fragments.

Additionally, recombinant anti-polymorphic protein antibodies, such as chimeric and humanized monoclonal antibodies, comprising both human and non-human portions, which can be made using standard recombinant DNA techniques, are within the scope of the invention. Such chimeric and humanized monoclonal antibodies can be produced by recombinant DNA techniques known in the art, for example using methods described in PCT International Application No. PCT/US86/02269; European Patent Application No. 184,187; European Patent Application No. 171,496; European Patent Application No. 173,494; PCT International Publication No. WO 86/01533; U.S. Pat. No. 4,816,567; European Patent

Application No. 125,023; Better et al. (1988) Science 240:1041-1043; Liu et al. (1987) PNAS 84:3439-3443; Liu et al. (1987) J Immunol. 139:3521-3526; Sun et al. (1987) PNAS 84:214-218; Nishimura et al. (1987) Cancer Res 47:999-1005; Wood et al. (1985) Nature 314:446-449; Shaw et al. (1988) J Natl Cancer Inst 80:1553-1559); Morrison(1985) Science 229:1202-1207; Oi et al. (1986) BioTechniques 4:214; U.S. Pat. No. 5,225,539; Jones et al. (1986) Nature 321:552-525; Verhoeyan et al. (1988) Science 239:1534; and Beidler et al. (1988) J Immunol 141:4053-4060.

In one embodiment, methodologies for the screening of antibodies that possess the desired specificity include, but are not limited to, enzyme-linked immunosorbent assay (ELISA) and other immunologically-mediated techniques known within the art.

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Anti-polymorphic protein antibodies may be used in methods known within the art relating to the detection, quantitation and/or cellular or tissue localization of a polymorphic protein (e.g., for use in measuring levels of the polymorphic protein within appropriate physiological samples, for use in diagnostic methods, for use in imaging the protein, and the like). In a given embodiment, antibodies for polymorphic proteins, or derivatives, fragments, analogs or homologs thereof, that contain the antibody-derived CDR, are utilized as pharmacologically-active compounds in therapeutic applications intended to treat a pathology in a subject that arises from the presence of the cSNP allele in the subject.

An anti-polymorphic protein antibody (e.g., monoclonal antibody) can be used to isolate polymorphic proteins by a variety of immunochemical techniques, such as immunoaffinity chromatography or immunoprecipitation. An anti-polymorphic protein antibody can facilitate the purification of natural polymorphic protein from cells and of recombinantly produced polymorphic proteins expressed in host cells. Moreover, an anti-polymorphic protein antibody can be used to detect polymorphic protein (e.g., in a cellular lysate or cell supernatant) in order to evaluate the abundance and pattern of expression of the polymorphic protein. Anti-polymorphic antibodies can be used diagnostically to monitor protein levels in tissue as part of a clinical testing procedure, e.g., to, for example, determine the efficacy of a given treatment regimen. Detection can be facilitated by coupling (i.e., physically linking) the antibody to a detectable substance. Examples of detectable substances include various enzymes, prosthetic groups, fluorescent materials, luminescent materials, bioluminescent materials, and radioactive materials. Examples of suitable enzymes include horseradish peroxidase, alkaline phosphatase,

-g a lactosidase, or acetylcholinesterase; examples of suitable prosthetic group complexes include streptavidin/biotin and avidin/biotin; examples of suitable fluorescent materials include umbelliferone, fluorescein, fluorescein isothiocyanate, rhodamine, dichlorotriazinylamine fluorescein, dansyl chloride or phycoerythrin; an example of a luminescent material includes luminol; examples of bioluminescent materials include luciferase, luciferin, and aequorin, and examples of suitable radioactive material include ¹²⁵I, ¹³¹I, ³⁵S or ³H.

Map location	61	m
Similiarity (pValue) following a BLASTX analysis	0	0
Protein Name of protein identified classification of following a BLASTX analysis of CuraGen gene the CuraGen sequence	ATPase_associat Human Gene SWISSFROT- ed D.:P20648 POTASSIUM- TRANSPORTING ATPASE ALPHA CHAIN (EC 3.6.1.36) (PROTON PUMP) (GASTRIC H+/K+ ATPASE ALPHA SUBUNIT) - HOMO SAPIENS (HUMAN), 1035 aa.	Human Gene SWISSPROT- ID:P18084 INTEGRIN BETA-5 SUBUNIT PRECURSOR - HOMO SAPIENS (HUMAN), 799 aa.
Protein classification of CuraGen gene	ATPase_associat	cadherin
Amino Amino Type of acid acid change before after	SILENT- CODING	SILENT: CODING
Amino acid after	Glu	ם
Amino acid before	Gln	Thr
Base after	<u> ප</u>	[-
Base before	⊀	4
Base pos. Polymorphic sequence Base of SNP	CGCTGACAGGGGA / GTCTGAGCCACA[A GJACCCGCTCACC CGAGTGCACGCAC G	ATGGATAGTCCAT CTGGTTGGATGC[A /I]GTGTACTCGTTG GCCTCGTTCAGGT
Base pos. of SNP	1008	2296
Seq ID CuraGen sequence ID	cg4333349	cg43931765
Seq ID	-	2

· =	<u>&</u>
o .	0
Human Gene SWISSNEW- ID:P13591 NEURAL CELL ADHESION MOLECULE, 140 KD ISOFORM PRECURSOR (N- CAM 140) (NCAM-140) (CD56 ANTIGEN) - HOMO SAPIENS (HUMAN), 848 aa.pcls:SWISSPROT-ID:P13591 NEURAL CELL ADHESION MOLECULE, 140 KD ISOFORM PRECURSOR (N-CAM 140) (NCAM-140) (CD56 ANTIGEN) - HOMO SAPIENS (HUMAN), 848 aa.	Human Gene SWISSNEW- D::Q08554 DESMOCOLLIN 1A/1B PRECURSOR (DESMOSOMAL (DESMOSOMAL (DGZ/DG3) - HOMO SAPIENS (HUMAN), 894 aa. pcis:SWISSPROT-ID::Q08554 DESMOCOLLIN 1A/1B PRECURSOR (DESMOSOMAL GLYCOPROTEIN 2/3) (DG2 / DG3) - HOMO SAPIENS (HUMAN), 894 aa.
cadherin	cadherin
SILENT	SILENT- CODING
Val	
Val	<u>#</u>
O .	U
AATACAAAGCTGA GTGGAGAGGAGTT /GJGGTGAAGAAGT ATGGCATTCCAAG T	TATTOTTATTATGT / ATTCTGTTTAC[A/G JTGTTTCTGTGTCA CTGCTAAGAGAA
1832	2330
cg44130533	cg34888922
m	4

	_		
82	17 (17q21.3 2)	17 (17q21.3 2)	17 (17q21.3 2)
0	0	0	0
Human Gene SWISSNEW- ID:Q08554 DESMOCOLLIN 1A/1B PRECURSOR (DESMOSOMAL GL YCOPROTEIN 2/3) (DG2/DG3) - HOMO SAPIENS (HUMAN), 894 aa,pcis:SWISSPROT-ID:Q08554 DESMOCOLLIN 1A/1B PRECURSOR (DESMOSOMAL GL YCOPROTEIN 2/3) (DG2 / DG3) - HOMO SAPIENS (HUMAN), 894 aa.	Human Gene SWISSPROT- ID:P08514 PLATELET MEMBRANE GLYCOPROTEIN IIB PRECURSOR (GPIIB) (INTEGRIN ALPHA- IIB) (CD41) - HOMO SAPIENS (HUMAN), 1039 aa.	Human Gene SWISSPROT- ID:P08514 PLATELET MEMBRANE GLYCOPROTEIN IIB PRECURSOR (GPIB) (INTEGRIN ALPHA- IIB) (CD41) - HOMO SAPIENS (HUMAN), 1039 aa.	Human Gene SWISSPROT- ID:P08514 PLATELET MEMBRANE GLYCOPROTEIN IIB PRECURSOR (GPIIB) (INTEGRIN ALPHA- IIB) (CD41) - HOMO SAPIENS (HUMAN), 1039 aa.
cadherin	cadherin	cadherin	cadherin
SILENT- CODING	SILENT- CODING	SILENT. CODING	SILENT: CODING
Tyr .	Val	Ala	Ser
Tyr	Val	Ala	Ser
[-4	ပ	O	D
O	5	ပ	U
CAAGGAGCATTGA CCGTGAGAAATA[C /I]GAACAGTTTGC GTTATATGGCTATG	TGGCGTCGTATTTT GGGCATTCAGT[G/ CJGCTGTCACTGAC GTCAACGGGGATG	AGGGGCCTATGA AGCAGAGCTGGC[C /GJGTGCACCTGCC CCAGGGCGCCCAC T	GTTACTGTGAAGC GGGCTTCAGCTC[C/ G GTGGTCACTCAG GCCGGAGAGCTGG
815	1172	2243	812
cg3488922			cg40310734 8
رم د	0	7	·

×	4	4	6 (6p21.3)
0	7.005-172	7.00E-172	8.00E-155
Human Gene SWISSPROT- ID:P32004 NEURAL CELL ADHESION MOLECULE L1 PRECURSOR (N-CAM L1) - HOMO SAPIENS (HUMAN), 1257 aa.	Human Gene SWISSPROT- ID:P21815 BONE SIALOPROTEIN II PRECURSOR (BSP II) (CELL-BINDING SIALOPROTEIN) (INTEGRIN- BINDING SIALOPROTEIN) - HOMO SAPIENS (HUMAN), 317 aa.	Human Gene SWISSPROT- ID:P21815 BONE SIALOPROTEIN II PRECURSOR (BSP II) (CELL-BINDING SIALOPROTEIN) (INTEGRIN- BINDING SIALOPROTEIN) - HOMO SAPIENS (HUMAN), 317 aa.	Human Gene SPTREMBL- ID:Q64411 PROGASTRICSIN PRECURSOR (EC 3.4.23.3) (PEPSIN C) - CAVIA PORCELLUS (GUINEA PIG), 394 aa.
cadherin	cadherin	cadherin	cathepsin
SILENT- CODING	SILENT. CODING	SILENT. CODING	SILENT: CODING
Gly	Asn	Asp	Leu
Gly	Asn	Asp	Leu
H	ļ-	<u>-</u>	G
5	U	U	U
GTGACAAGTACTT CATAGAGGATGG[GT]CGCCTGGTCAT CCACAGCCTGGAC T	AAGGAGAAAACAA TGAAGAACCGAA[CT]GAAGACGAAG ACTCTGAGGCTGA GA	AAAACAATGAAGA ACCGAACGAAGA[CT]GAAGACTCTG AGCTGAGAATAC CA	AGAACGGCCAGCC CCTGTGGATCCT[C/ GJGGGGATGTCTTC CTCAGGTCCTACT
1922	383	389	1289
cg43331935	cg42388009	cg42388009	cg44126574
٥	01		

3 (3p21.3)	1 (1p32)	9 (9q34.3)	1 (1q32)	1 (1p21)
0	0	1.40E-104	1.10E-69	5.00E-304
Human Gene SWISSPROT- ID:Q02388 COLLAGEN ALPHA I(VII) CHAIN PRECURSOR (LONG-CHAIN COLLAGEN) (LC COLLAGEN) - HOMO SAPIENS (HUMAN), 2944 aa.	Human Gene SWISSPROT- ID:P07357 COMPLEMENT COMPONENT C8 ALPHA CHAIN PRECURSOR - HOMO SAPIENS (HUMAN), 584 aa.	Human Gene Homologous to SWISSPROT-ID:P07360 COMPLEMENT C8 GAMMA CHAIN PRECURSOR - HOMO SAPIENS (HUMAN), 202 aa.	Human Gene Similar to TREMBLNEW-ID:E246058 COMPLEMENT RECEPTOR 2 - MUS MUSCULUS (MOUSE), 651 aa (fragment).	Human Gene SWISSPROT- ID:P09603 MACROPHAGE COLONY STIMULATING FACTOR-1 PRECURSOR (CSF-1) (MCSF) - HOMO SAPIENS (HUMAN), 554 aa.
collagen	complement	complement	pt pt	csf
SILENT. CODING	SILENT- CODING	SILENT- CODING	SILENT: CODING	SILENT- CODING
Ser	Ala	Ala	Gly	Thr
Ser	Ala	Ala	Gly	Th
H	_ව	ى ت	0	ပ
O	∢	H	∢	∢ .
GGACTCCAGTGTC CAGGGCATCCAG[C /IJTACATCCTATCC TGGCGGCCACTCA	TAAGACGGGCAGC TACACCCGCAGCIA /GJGTTACCTGCCA GCTGAGCAACTGG T	TCCAGCCCAAGGC CAATTTTGATGC[T/ G]CAGCAGTTTGCA GGGACCTGGCTCC	AGGTAGGAGGGCT TGGTCTCCAAACIA /GJCCTATTGTTTCA TTCTCCACAGTGC	AACAGCCGCCGCAGA TGTAACTGGTACIA /CJGCCTTGCCCAG GGTGGGCCCCGTG A
3066	245	222	1371	1219
cg43970983	cg44032748	cg41553795	cg43942011	cg2164442
E1	14	15		17

17 (17q11.2)		(10925)
1.50E-107 ((1.60E-266	2.00E-229 10 (10
Human Gene Homologous to SWISSPROT-ID:P09919 GRANULOCYTE COLONY- STIMULATING FACTOR PRECURSOR (G-CSF) (PLURIPOIETIN) - HOMO SAPIENS (HUMAN), 207 aa.	Human Gene TREMBLNEW- ID:G2979625 PYRUVATE DEHYDROGENASE COMPLEX PROTEIN X SUBUNIT PRECURSOR - HOMO SAPIENS (HUMAN), 501 aa.	Human Gene SWISSPROT- ID:P45954 ACYL-COA DEHYDROGENASE, SHORTBRANCHED CHAIN SPECIFIC PRECURSOR (EC 1.3.99) (SBCAD) (2-METHYL DEHYDROGENASE) (2- MEBCAD) - HOMO SAPIENS (HUMAN), 432 aa.
ÇSÎ.	dehydrogenase	dehydrogenase
SILENT.	SILENT- CODING	SILENT- CODING
Leu	Pro	Ser
	o O	Ser
<	Ö	F
ე	∢	
TCCAGCGCCGGGC AGAGGGGTCCT[G AJGTTGCCTCCCAT CTGCAGAGCTTCC	ATGTGCCCACTGC ATTGGGTTGTCC[A/ GJGGAGTTGATACT GGTGGGATCACAG	CAGAATATGGAGG CACAGGAGCTTC[A /IJTTTTTATCCACT GTGCTCGTGATAG
597	1743	366
cg41533258	cg43996714	cg43259523
<u>&</u>	61	20

4 (49,22)			
1.30E-209	2.50E-103	2.50E-103	2.50E-103
Human Gene SWISSNEW- ID:P08319 ALCOHOL DEHYDROGENASE CLASS II PI CHAIN (EC 1.1.1) - HOMO SAPIENS (HUMAN), 391 aa.lpcls:SWISSPROT-ID:P08319 ALCOHOL DEHYDROGENASE CLASS II PI CHAIN (EC 1.1.1.1) - HOMO SAPIENS (HUMAN), 391	Human Gene Homologous to SPTREMBL-ID:Q92816 CYTOPLASMIC DYNEIN 3 HEAVY CHAIN - HOMO SAPIENS (HUMAN), 197 aa (fragment).	Human Gene Homologous to SPTREMBL-ID:Q92816 CYTOPLASMIC DYNEIN 3 HEAVY CHAIN - HOMO SAPIENS (HUMAN), 197 aa (fragment).	Human Gene Homologous to SPTREMBL-ID:Q92816 CYTOPLASMIC DYNEIN 3 HEAVY CHAIN - HOMO SAPIENS (HUMAN), 197 aa (fragment).
dehydrogenase	dynein	dynein	dynein
SILENT - CODING	SILENT- CODING	SILENT. CODING	SILENT- CODING
Leu	Thr	Lys	Leu
Leu	Thr	Lys	Leu
E	ව	ව	<u>F</u>
ပ	V	, 4	U
GAATAAGAAATTC AATCTGGATGCA[C /TJTGGTGACCCATA CCCTGCCTTTTGA	GTGCTCCAGAGGG GCCAGCAGGCAC[A/G]GGAAAAACCG AAACCACCAAGGA CT	CAGAGGGGCCAGC AGGCACAGGAAA[A/G]ACCGAAACCA CCAAGGACTTGGC TA	AGCAATGGGAAAG TTTTTAAAGGA[C/ TJTGGCTTCTTCTG GTGCTTGGGCTTG
1528	430	436	542
cg43057018	cg1395871	cg1395871	cg1395871
21	22	23	24

	16	ა
2.50E-103	0	1.00E-104
Human Gene Homologous to SPTREMBL-ID:Q92816 CYTOPLASMIC DYNEIN 3 HEAVY CHAIN - HOMO SAPIENS (HUMAN), 197 aa (fragment).	Human Gene TREMBLNEW- ID: G2865466 HEAT SHOCK PROTEIN 75 - HOMO SAPIENS (HUMAN), 649 aa.	Human Gene Homologous to SWISSNEW-ID:Q52500 THERMOSOME SUBUNIT (HEAT-SHOCK PROTEIN) - PYROCOCCUS KODAKARAENSIS, 546 aa.lpcls:SWI§SPROT-ID:Q52500 THERMOSOME SUBUNIT (HEAT-SHOCK PROTEIN) - PYROCOCCUS SP. (STRAIN KODI), 546 aa.
dynein	eph	eph
SILENT- dynein CODING	SILENT: CODING	SILENT- CODING
Phe	Asp	Arg
Phe	Asp	Arg
[V	L
ပ	ပ	O
CTTCTTCTGGTGCT C TGGGCTTGCTT[C/T]GATGAATTCAACC GGATTGAGTTGG	AGCGGCCCACCAT GGCCTAGGGTC[G /AJTCAACAAGTCC AGCAGCAATCATG G	CCGATGGCTATGA GCAGGCTGCTCG[C //]GTTGCTATTGAA CACCTGGACAAGA
571	1269	461
cg1395871	cg43950268	cg43918531
25	76	27

1.90E-178	
Human Gene SWISSNEW- Di.Q15166 SERUM PARAOXONASE/ARYLESTERA SE 3 (EC 3.1.1.2) (EC 3.1.8.1) (PON 3) (SHRUM ARYLDIAKYLPHOSPHATASE 3) (A-ESTERASE 3) (AROMATIC ESTERASE 3) (AROMATIC ESTERASE 3) HOMO SAPIENS (HUMAN), 341 aa (fragment).[pcis:SWISSPROT- ID:Q15166 SERUM PARAOXONASE/ARYLESTERA SE 3 (EC 3.1.1.2) (EC 3.1.8.1) (PON 3) (SERUM ARYLLIZAKYLPHOSPHATASE 3) (A-ESTERASE 3) (AROMATIC ESTERASE 3) (AROMATIC ESTERASE 3) HOMO SAPIENS (HUMAN), 341 aa (fragment).	
cstcrase	
SILENT- CODING	
DIO.	
Qin	
H	
υ.	
CAAGITCCTCAAT AAAGIGGCAGIT(C TJTCAGGITCTACT GGCTCCACTTCTC	
cg43957743 1146	

	5 (5q34)	5 (5q34)
3.30E-60	1.90E-256 5	1.30E-248 5
Human Gene Similar to SWISSNEW-ID:Q23917 3;5- CYCLIC-NUCLEOTIDE PHOSPHODIESTERASE REGA (EC 3.1.4.17) (PDEASE REGA)- DICTYOSTELJUM T93 aa,pcls:SWISSPROT- ID:Q23917 3;5-CYCLIC- NUCLEOTIDE PHOSPHODIESTERASE REGA (EC 3.1.4.17) (PDEASE REGA)- DICTYOSTELJUM DISCOIDEUM (SLIME MOLD), 793 aa.	Human Gene SWISSPROT- ID:P47870 GAMMA- AMINOBUTYRIC-ACID RECEPTOR BETA-2 SUBUNIT PRECURSOR (GABA(A) RECEPTOR) - HOMO SAPIENS (HUMAN), 474 aa.	Human Gene SWISSPROT- ID:P14867 GAMMA- AMINOBUTYRIC-ACID RECEPTOR ALPHA-1 SUBUNIT PRECURSOR (GABA(A) RECEPTOR) - HOMO SAPIENS (HUMAN), 456 aa.
esterase	gaba	gaba
SILENT.	SILENT: CODING	SILENT- CODING
s V	Tyr	Gly
స్	Tyr	Gly
[-	<u></u>	H
O	O	U
TCACCCTCAGGAG GTGGCTGTTCTG[C/ TJGTCCACGACAAC TACAGAAACAACC	TCTTCAACATGGTC TATTGGCTTTA[C/T JTATGTGAACTAAA ACATGGCCTCCC	GGATTTGGACAG ACTCCTAGATGG[C //JTATGACAATCGC CTGAGACCAGGAT
5963	1631	370
cg43319420	cg3001932	cg43975899
55	30	E .

17 (17q25.2)	17 (17q25.2)	7 (7q21.11)	8	15 (15q15)
7.40E-199	7.40E-199	,	7.40E-80	0
Human Gene TREMBL/NEW-D:G2826521 MALTASE-GLUCOAMYLASE (EC 3.2.1.20) - HOMO SAPIENS (HUMAN), 1857 aa.	Human Gene TREMBL/NEW- ID:G282652 MALTASE- GLUCOAMYLASE (EC 3.2.1.20) - HOMO SAPIENS (HUMAN), 1857 aa.	Human Gene SWISSPROT- ID:P08236 BETA- GLUCURONIDASE PRECURSOR (EC 3.2.1.31) (BETA-G1) - HOMO SAPIENS (HUMAN), 651 aa.	Human Gene Similar to SWISSPROT-ID:P08236 BETA- GLUCURONIDASE PRECURSOR (EC 3.2.1.31) (BETA-G1) - HOMO SAPIENS (HUMAN), 651 aa.	Human Gene SWISSPROT- ID:P16452 ERYTHROCYTE MEMBRANE PROTEIN BAND 4.2 (P4.2) (PALLIDIN) - HOMO SAPIENS (HUMAN), 690 aa.
glucoamylase	glucoamylase	glucuronidase	glucuronidase	glycoprotein
SILENT- CODING	SILENT- CODING	SILENT- CODING	SILENT- CODING	SILENT- CODING
Gln	Arg	Туг	Tyr	Ala
Gln	Arg	Tyr	Tyr	Ala
<u>0</u>	∢	₹	∢	A
∀	ق ق	o	, ප	5
GGGCCCACTTCCC CCTGGACGTCCAIA /GJTGGAACGACCT GGACTACATGGAC T	TGAACGAGCCTTC CAACTTCATCAGG AJGGCTCTGAGGA CGGCTGCCCCAAC A	AATTCCAAATGAG CTCTCCAACCAC[G/ AJTATTTCTGCGT TTTTGATCCAGAC	AATTCCAGATGAG CTCTCCAACCAC[G/ AJTATTTTCTGCGT TTTTGATCCAGAC	GGACCATCTCTGT GACCACACCTGC[G /A]GACGCTGTCATT GGCCACTACTCGC
1643	2021	443	325	088
cg43299024		cg43969076		cg43065549
32	33	34	35	36

15 (15q15)			X (Xq28)
0	0	0	3.10E-249
Human Gene SWISSPROT- ID:P16452 ERYTHROCYTE MEMBRANE PROTEIN BAND 4.2 (P4.2) (PALLIDIN) - HOMO SAPIENS (HUMAN), 690 aa.	Human Gene SWISSPROT- D:Q12889 OVIDUCT-SPECIFIC GLYCOPROTEIN PRECURSOR (OVIDUCTIN) GLYCOPROTEIN) (OVIDUCTIN) (ESTROGEN- DEPENDENT OVIDUCT PROTEIN) - HOMO SAPIENS (HUMAN), 678 aa.	Human Gene SWISSPROT- ID:Q12889 OVIDUCT-SPECIFIC GLYCOPROTEIN PRECURSOR (OVIDUCTAL GLYCOPROTEIN) (OVIDUCTIN) (ESTROGEN- DEPENDENT OVIDUCT PROTEIN) - HOMO SAPIENS (HUMAN), 678 aa.	Human Gend SWISSPROT- ID:Q00013 55 KD ERYTHROCYTE MEMBRANE PROTEIN (P55) - HOMO SAPIENS (HUMAN), 466 aa.
glycoprotein	glycoprotein	glycoprotein	głycoprotein
SILENT. CODING	SILENT- CODING	SILENT- CODING	SILENT- CODING
Val	Val	Lys	Thr
Val	Val	Lys	Thr
9	E	[H	υ
<u></u>	U	0	∀
ACCCTGGAATAG AGAGGATGCTGTIT /GJTTCCTGAAGAA TGAGGCTCAGCGC A	TGACGTCATCCAT GTCCAATGTCCA[C/ TJACCATGGCCCC CCAAAATGCTCTC	GAAGGGATATAAC TGAAGCAATAAA[C //]TTTCACGGTTG GCAAATGTGGACA	AGGACTGTTTTTCA TTCAGCTTCAG[AC JGTGATTCCCATGG GCTCTTCTGTGA
166	1141	1846	1677
cg43065549	cg44004239	cg44004239	cg43957605
37	38	39	40

1 (1921)	1 (1921)
2.00E-183	6.70E-183
Human Gene SWISSNEW- ID:P06126 T-CELL SURFACE GLYCOPROTEIN CDIA PRECURSOR (CDIA ANTIGEN) (T-CELL SURFACE ANTIGEN T6/LEU-6) (HTA1 THYMOCYTE ANTIGEN) - HOMO SAPIENS (HUMAN), 327 aa.jpcls:SWISSPROT-ID:P06126 T-CELL SURFACE GLYCOPROTEIN CDIA PRECURSOR (CDIA ANTIGEN) (T-CELL SURFACE ANTIGEN) (T-CELL SURFACE ANTIGEN) T6/LEU-6) (HTA1 THYMOCYTE	Human Gene SWISSNEW- D:P29016 T-CELL SURFACE GLYCOPROTEIN CD1B ACTORNOR (CD1B ANTIGEN) - HOMO SAPIENS (HUMAN), 333 aa_[pcls:SWISSPROT- D:P29016 T-CELL SURFACE GLYCOPROTEIN CD1B PRECURSOR (CD1B ANTIGEN) - HOMO SAPIENS (HUMAN), 333 aa_
glycoprotein	glycoprotein
SILENT: CODING	SILENT.
Pro	Leu
Pro	Leu
[H
O	O
ATGTCTCAGGATTC TACCCAAGCC[C/ TGTGTGGGTGATG TGGATGCGGGTG	TGGCAATAATAGT GCCTTCCTTGCT[C/ TJCTTTTGCTATGC CTTGCATTATGGT
1229	1210
	cg40356255
[4	

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	1 (1921)	14 (14q11.2')
7.60E-127	1.60E-119	9.50E-70
Human Gene Homologous to SWISSNEW-ID:P01732 T-CELL SURFACE GLYCOPROTEIN CD8 ALPHA CHAIN PRECURSOR (T-LYMPHOCYTE DEFERENTIATION ANTIGEN 78/LEU-2) - HOMO SAPIENS (HUMAN), 235 aa. pcls:SWISSPROT-ID:P01732 T-CELL SURFACE GLYCOPROTEIN CD8 ALPHA CHAIN PRECURSOR (T- LYMPHOCYTE GLYCOPROTEIN CD8 CHAIN PRECURSOR (T- LYMPHOCYTE TSMERENTIATION ANTIGEN T8/LEU-2) - HOMO SAPIENS (HUMAN), 235 aa.	Human Gene Homologous to SWISSPROT-ID:P02743 SERUM AMYLOID P-COMPONENT PRECURSOR (SAP) (9.5S ALPHA-1-GLYCOPROTEIN) - HOMO SAPIENS (HUMAN), 223 aa.	Human Gene Similar to SWISSPROT-ID:P16452 ERYTHROCYTE MEMBRANE PROTEIN BAND 4.2 (P4.2) (PALLIDIN) - HOMO SAPIENS (HUMAN), 690 aa.
glycoprotein	glycoprotein	glycoprotein
CODING	SILENT: CODING	SILENT- CODING
Leu	Val	Gly
Leu	Val	Gly
Ę-	∢	∢
lo lo	o .	υ
CTGTGATATCTACA TCTGGGCGCCC[CT JTGGCCGGGACTTG TGGGGTCCTTCT	AGGGTCTGCGACA GGGTTACTTTGT[G/ A]GAAGCTCAGCCC AAGATTGTCCTGG	ATGGCCAGTGCTG GGTCTTTGCTGG[C/ AJGTGACCACCACA GTGCTGCGCTGCC
. 1183	544	1242
cg44004667	cg43068999	cg41568631
	44	45

14 (14q11.2)	1 (1922)	1 (1922)	17	
9.90E-70	3.00E-52	3.00E-52	3.60E-120	4.30E-216
Human Gene Similar to SWISSPROT-ID:P16452 ERYTHROCYTE MEMBRANE PROTEIN BAND 4.2 (P4.2) (PALLIDIN) - HOMO SAPIENS (HUMAN), 690 aa.	Human Gene Similar to SPTREMBL-ID:Q91406 IP1=CNS MYELIN P0-LIKE GLYCOPROTEIN - UNKNOWN, 202 aa.	Human Gene Similar to SPTREMBL-ID:Q91406 IP1=CNS MYELIN P0-LIKE GLYCOPROTEIN - UNKNOWN, 202 as.	Human Gene Homologous to SWISSPROT-ID:Q12099 PROBABLE ATP-DEPENDENT RNA HELICASE FAL1 - SACCHAROMYCES CEREVISIAE (BAKER'S YEAST), 399 aa.	Human Gene SWISSPROT- ID:PS0458 HOMEOBOX PROTEIN LH-2 - HOMO SAPIENS (HUMAN), 423 aa.
glycoprotein	glycoprotein	glycoprotein	helicase	homeobox
SILENT- CODING	SILENT: CODING	SILENT: CODING	SILENT- CODING	SILENT: CODING
Gly	Arg	Asn	Ala	Asn
Gly	Arg	Asn	Ala	Asm
ე	<u> </u>	ပ	ပ	⊢
O	ပ	-	O	ပ
GCTCTGTGGAGTC CATCAAGAATGG[C /G]CTGGTCTACATG AAGTACGACACGC	GCATCCAGTGGGT AGGGGACCCTCG[C TJTGGAAGGATGG CTCCATTGTCATAC	TACACAACCTAGA CTACAGTGACAA[T /C]GGCACGTTCACT TGTGACGTCAAAA	AGTCCCTTCTCCGT GGCACCTACGC[G/ CJTATGGTTTTGAG AAGCCCTCTGCCA	AGTCTTACTTTGCC ATTAACCACAA[C/ TJCCCGACGCCAAG GACTTGAAGCAGC
1545	361	409	465	1353
cg41568631		cg41603916		cg43983917
46	47	48	49	20

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				2 (2421)
9	2	£.	E	2 (
4.30E-216	2.60E-188	1.10E-123	1.30E-113	0
Human Gene SWISSPROT- ID:P50458 HOMEOBOX PROTEIN LH-2 - HOMO SAPIENS (HUMAN), 423 aa.	Human Gene SWISSPROT- ID:P28356 HOMEOBOX PROTEIN HOX-D9 (HOX-4C) (HOX-5.2) - HOMO SAPIENS (HUMAN), 342 aa.	Human Gene Homologous to SWISSPROT-ID:P17509 HOMEOBOX PROTEIN HOX-B6 (HOX-2B) (HOX-2.2) (HU-2) - HOMO SAPIENS (HUMAN), 224 aa.	Human Gene Homologous to SWISSPROT-ID:P09629 HOMEOBOX PROTEIN HOX-B7 (HOX-2C) (HHO.C1) - HOMO SAPIENS (HUMAN), 217 aa.	Human Gene SWISSPROT- ID:P09848 LACTASE- PHLORIZIN HYDROLASE PRECURSOR (EC 3.2.1.108) (EC 3.2.1.62) (LACTASE- GLYCOSYLCERAMIDASE) - HOMO SAPIENS (HUMAN),
homeobox	нотеорох	homeobox	homeobox	hydrolase
SILENT- CODING	SILENT- CODING	SILENT- CODING	SILENT- CODING	SILENT:- CODING
Asp	Ala	Lys	Lys	Arg
Asp	Ala	Lys	Lys	Arg
<u>_</u>	£	∢	ပ	ර
ပ	9	ච	[V
ACTTTGCCATTAAC CACAACCCCGA[C/ TJGCCAAGGACTTG AAGCAGCTCGCGC	TGGAGCGAGCGTG GATCCAGTTCGC[G //JGCGGGGTTGTTT GGGTCAAGTTGCT	GTTACCAGACGCT GGAGCTGGAGAA[G/A]GAGTTTCACT ACAATCGCTACCT GA	TCAGGTAGCGATT GTAGTGAAATTC[T/ CJTTCTCCAGCTCC AGGGTCTGGTAGC	GGGAAGCATITGC CAATCAGTCCAGIA /GJGCGGAAAGGA TGCCTTCCTGCAGG
1359	979	689	810	1124
cg43983917	cg42730678	cg42714160	cg43959084	cg42359655
51	52	53	54	

2 (2q21)	2 (2q21)	16 (16q22)	15
0	0	2.00 <u>5</u> -220	0
Human Gene SWISSPROT- ID:P09848 LACTASE- PHI,ORIZIN HYDROLASE PRECURSOR (EC 3.2.1.108) (EC 3.2.1.62) (LACTASE- GLYCOSYLCERAMIDASE)- HOMO SAPIENS (HUMAN), 1927 aa.	Human Gene SWISSPROT- ID:P09848 LACTASE- PHLORIZIN HYDROLASE PRECURSOR (EC 3.2.1.108) (EC 3.2.1.62) (LACTASE- GLYCOSYLCERAMIDASE) - HOMO SAPIENS (HUMAN),	Human Gene SPTREMBL- ID:Q13194 11-BETA- HYDROXYSTEROID DEHYDROGENASE TYPE 2 - HOMO SAPIENS (HUMAN), 405 aa.	Human Gene TREMBLNEW- ID:G2114410 INTERLEUKIN-16 - HOMO SAPIENS (HUMAN), 631 aa.
hydrolase	hydrolase	hydroxysteroid	interleukin
SILENT. CODING	SILENT- CODING	SILENT: CODING	SILENT. CODING
His	Gly	Val	Pro
His	Gly		Pro
[-	E	∢	H
O	O	U	U
ACAGCCAGCGGTT O TGGCCTGCACCA[C //JGTCAACTTCAGC GACAGCAGCAAGT	ATCTGGTCACCCTG C CAGAACCTGGG C/ TIGTGTCCCACTAC CGTTTTCCATCT	TGGTGTGGGCCTT GGTGAACTCTAG[C /A]ACGCGGCTAAT GTCTCCTGGTTTGG	GGAAGCTGACTCC (AGAGGCCATGCCIC //IGACCTCAACTCC TCCACTGACTCTG
2468	4340	1329	1689
	cg42359655		cg43922672
36	57		59

7 (7p21)	5 (5p13)
3.40E-108	3.10E-249
Human Gene Homologous to SWISSPROT-ID:P05231 INTERLEUKIN-6 PRECURSOR (IL-6) (B-CELL STIMULATORY FACTOR 2) (BSF-2) (INTERFERON BETA-2) (HYBRIDOMA GROWTH FACTOR) - HOMO SAPIENS (HUMAN), 212 aa.	interleukinrecept Human Gene SWISSNEW- ID:P16871 INTERLEUKIN-7 RECEPTOR ALPHA CHAIN RECEPTOR ALPHA CHAIN PRECURSOR (IL-7R-ALPHA) (CDW127) (CD127 ANTIGEN) - HOMO SAPIENS (HUMAN), 459 aa.pcls:SWISSPROT-ID:P16871 INTERLEUKIN-7 RECEPTOR ALPHA CHAIN PRECURSOR (IL-7R-ALPHA) (CDW127) (CD127 ANTIGEN) - HOMO SAPIENS (HUMAN), 459 aa.
interleukin	interleukinrecept
SILENT- CODING	SILENT. CODING
Val	His
	His
U_	1
D	U
GTAGTGAGGAACA C AGCCAGAGCTGT[G /C]CAGATGAGTAC AAAAGTCCTGATC C	AGTTGGAAGTGAA TGGATCGCAGCA[C AJTCACTGACCTGT GCTTTTGAGGACC
	181
cg42908571 630	cg43942050
09	61

		r
E.	6 _	01
·		
0	0	0
Human Gene SWISSNEW- ID:P42336 PHOSPHATIDYLINOSITOL 3- KINASE CATALYTIC SUBUNIT, ALPHA ISOFORM (EC 2.7.1.137) (P13-KINASE P110 SUBUNIT ALPHA) (PTDINS-3- KINASE P110) (P13K) - HOMO SAPIENS (HUMAN), 1068 aa.lpcis:SWISSPROT-ID:P42336 PHOSPHATIDYLINOSITOL 3- KINASE CATALYTIC SUBUNIT, ALPHA ISOFORM (EC 2.7.1.137) (P13K) - HOMO SUBUNIT ALPHA) (PTDINS-3- KINASE P110) (P13K) - HOMO SUBUNIT ALPHA) (PTDINS-3- KINASE P110) (P13K) - HOMO SAPIENS (HUMAN), 1068 aa.	Human Gene SPTREMBL- ID:Q63553 SNF1-RELATED KINASE - RATTUS NORVECICUS (RAT), 746 aa.	Human Gene SWISSPROT- ID:Q04759 PROTEIN KINASE C, THETA TYPE (EC 2.7.1) (NPKC-THETA) - HOMO SAPIENS (HUMAN), 706 aa.
kinase	kinasc	kinase
SILENT CODING	SILENT: CODING	SILENT: CODING
	Asp	Phe
116	Asp	Phe
[E-I	l-	H
O	ပ	O
TAAATATTCGAGA CATTGACAAGATIC MJTATGTTCGAACA GGTATCTACCATG	AGATCTTTGAGGA AGGGGAATCTGA[C /T]GATGAGTTTGAC ATGGATGAGAATC	TTCTGACGCACAT GTTTTGTACATT[C/ T]CAGACCAAGGA AAACCTCTTTTTG
1249	1693	1438
cg43145505		cg43090990
	63	64

21 (21922.1)	X (Хq21.3)	1 (1421)
0	0	9.805-308
Human Gene SWISSPROT- ID:Q13627 SERINE/THREONINE-SPECIFIC PROTEIN KINASE MINIBRAIN HOMOLOG (EC 2.7.1) (HP86) (DYRK) - HOMO SAPIENS (HUMAN), 763 aa.	Human Gene SWISSPROT- ID:Q06187 TYROSINE-PROTEIN KINASE BTK (EC 2.7.1.112) (BRUTON'S TYROSINE KINASE) (AGAMMAGLOBUL.NAEMIA TYROSINE KINASE) (TYROSINE KINASE) (EPK) - HOMO SAPIENS (HUMAN), 659 aa.	Human Gene SPTREMBL- ID:Q16715 PYRUVATE KINASE (EC 2.7.1.40) - HOMO SAPIENS (HUMAN), 387 aa (fragment).
kinase	kinase	kinase
SILENT- kinase CODING	CODING	SILENT- CODING
Lys	Cys	Arg
Lys	Cys	Arg
<u>.</u>	ပ	U .
	T	4
TTAGTATCATTCAC A TGTGATCTAAA[A/ GJCCTGAAAATATC CTTCTTTGTAACC	AGGTATATACCAT CATGTACAGTTG[T/ CJTGGCATGAGAAA GCAGATGAGCGTC	TOGCTCCGGCTAC / ACCAACATCATG[A /C]GGGTGCTAAGC /ATATCCTGAGACG C
	2062	1744
cg43969763 2339	cg42879455	cg42659872
59	99	

7 (7q21.3)	17	11 (20p13)
1.60E-220	3.80E-219	2.00E-215
Human Gene SWISSPROT- DD:Q16654 [PYRUVATE DEHYDROGENASE(LIPOAMID E)] KİNASE ISOZYME 4 PRECURSOR (EC 2.7.1.99) (FYRUVATE DEHYDROGENASE KINASE ISOFORM 4) - HOMO SAPIENS (HUMAN), 411 aa.,pcls:SPTREMBL-ID:Q16654 PYRUVATE DEHYDROGENASE KINASE ISOFORM 4 - HOMO SAPIENS (HUMAN), 411 aa.	Human Gene SWISSPROT- D:Q15119 [PYRUVATE DEHYDROGENASEQLIPOAMID E]) KINASE ISOZYME 2 PRECURSOR (EC 2.7.1.99) (PYRUVATE DEHYDROGENASE KINASE ISOFORM 2) - HOMO SAPIENS (HUMAN), 407 aa.jpcis:SPTREMBL-ID:Q15119 PYRUVATE DEHYDROGENASE KINASE - HOMO SAPIENS (HUMAN), 407 aa.	Human Gene SWISSPROT- ID:P19138 CASEIN KINASE II, ALPHA CHAIN (CK II) (EC 2.7.1.37) - HOMO SAPIENS (HUMAN), AND BOS TAURUS (BOVINE), 391 aa.
kinase	kinase	kinase
SILENT- CODING	CODING	SILENT- CODING
Ala	Arg	Ti Ti
Ala	Arg	Thr
ပ	<u></u>	<u></u>
₹	U	U
GCTTGCCAATTTCT CGTCTGTATGC[A/C JAAGTACTTTCAAG GAGATCTGAATC	CTGTGGGGTACAT GTAGCTGAAGAGIC MJCGCTCAATCTTC CTCAAGGGAACAC	ACATCATATTGGC GCTGCTGACGGG[C //IGTACTGCCCCCT GGCATGCTAGATG
1323	526	1448
		cg43917871
89	69	70

11 (20p13)	91	1	2
2.00E-215	7.80E-173	2.10E-154	7.90E-283
Human Gene SWISSPROT- ID:P19138 CASEIN KINASE II, ALPHA CHAIN (CK II) (EC 2.7.1.37) - HOMO SAPIENS (HUMAN), AND BOS TAURUS (BOVINE), 391 aa.	Human Gene SPTREMBL- ID:Q15599 TYROSINE KINASE ACTIVATOR PROTEIN I (TKA- 1) - HOMO SAPIENS (HUMAN), 450 aa.	Human Gene SPTREMBL- ID:Q27467 SIMILARITY TO TYROSINE-PROTEIN KINASE - CAENORHABDITIS ELEGANS, 1280 aa.	Human Gene SWISSPROT- ID:Q04771 ACTIVIN RECEPTOR TYPE I PRECURSOR (EC 2.7.1) (ACTR-I) (SERINE/THREONINE-PROTEIN (SERINE/THREONINE-PROTEIN (ACTIVIN RECEPTOR LI) (SKR1) (ACTIVIN RECEPTOR-LIKE KINASE 2) (ALK-2) (TGF-B SUPERFAMIL Y RECEPTOR TYPE I) (TSR-I) - HOMO SAPIENS (HUMAN), 509 aa.
kinase	kinase	kinase	kinasereceptor
SILENT- CODING	SILENT: CODING	SILENT: CODING	SILENT- CODING
Ala		Asn	Ala
Ala	S S S	Asn	Ala
5	<	U	ပ
<u>-</u>	O	j	E
CAGTGTAGAAATA GGGGTGCTCCATIT/ GJGCCTCTCTTGCA GTAAGCCGTGACT	AGCTCAATGGTGG CTCTGCGTGCTC[G/ AJTCCCGAAGTGAC CTGCCTGGTTCCG	AATTCAACCCACT CATCTATGGCAA[IV CJGATGTGGATTCT GTGGATGTTGCAA	AGACCCGCCGTC CCCTGGCCAAGCIT /CJGTGGAGTGCTG CCAAGGGGACTGG T
1526	912	1765	
cg43917871	cg44131752	cg43969473	cg44025829
71	72	73	74

	,	T	T
6 (6p21.3)	6 (6p21.3)	6 (6p21.3)	6 (6p21.3)
9.10E-147	3.70E-134	3.70E-134	3.70E-134
Human Gene Homologous to SWISSPROT-ID:P28068 CLASS II HISTOCOMPATIBILITY ANTIGEN, M BETA CHAIN PRECURSOR - HOMO SAPIENS (HUMAN), 263 aa.	Human Gene Homologous to SWISSPROT-ID:P06340 HLA CLASS II HISTOCOMPATIBILITY ANTIGEN, DZ ALPHA CHAIN PRECURSOR (MHC DN-ALPHA) - HOMO SAPIENS (HUMAN), 250 aa.	Human Gene Homologous to SWISSPROT-ID:P06340 HLA CLASS II HISTOCOMPATIBILITY ANTIGEN, DZ ALPHA CHAIN PRECURSOR (MHC DN-ALPHA) - HOMO SAPIENS (HUMAN), 250 aa.	Human Gene Homologous to SWISSPROT-ID:P06340 HLA CLASS II HISTOCOMPATIBILITY ANTIGEN, DZ ALPHA CHAIN PRECURSOR (MHC DN-ALPHA) - HOMO SAPIENS (HUMAN), 250 aa.
МНС	MHC	мнс	МНС
SILENT- CODING	SILENT- CODING	SILENT- CODING	SILENT: CODING
Lea	Leu	Leu	Leu
Leu	Leu	Leu	Leu
₹	<u>.</u>	Y	A
g	Ą		් ව
TTAACACGAGGA GCCTGTGATGCT[G/ AJGCCTGCTATGTG TGGGGCTTCTATC	CCCCTGTGATCAAT ATCACCTGGCT[A/ GJCGCAACGGCCA AACTGTCACTGAG G	CACCACCAGATGC CATGGAGACCT[G /A]GTCTGTGCCATG GGCCTGGCCATCG	CCATGGAGACCCT GGTCTGTGCCCT[G/ AJGGCCTGGCCATC GGCCTGGTGGGCT
632		857	698
			cg42686658 8
76	77	78	67
	cg43960144 632 11AACACUGAGGA G A Leu Leu SILENT- MHC Human Gene Homologous to 9.10E-147 GCCTGTGATGCT[G/ A]GCCTGCTATGTG TGGGGCTTCTATC TGGGGCTTCTATC TGGGGCTTCTATC TGGGGCTTCTATC Human Gene Homologous to 9.10E-147 SWISSPROT-ID:P28068 CLASS II HISTOCOMPATIBILITY ANTIGEN, M BETA CHAIN PRECURSOR - HOMO SAPIENS (HUMAN), 263 aa.	CODING CONTINUE CODING CODING	GCCTGGCCTCGCTGGT A

				 1
6 (6p21.3)	6 (6p21.3)	6 (6p21.3)	19	61
3.70E-134	3.70E-134	3.70E-134	1.80E-113	1.80E-113
Human Gene Homologous to SWISSPROT-ID:P06340 HLA CLASS II HISTOCOMPATIBILITY ANTIGEN, DZ ALPHA CHAIN PRECURSOR (MHC DN-ALPHA) - HOMO SAPIENS (HUMAN), 250 aa.	Human Gene Homologous to SWISSPROT-ID:P06340 HLA CLASS II HISTOCOMPATIBILITY ANTIGEN, \$\tilde{p}\tilde{z}\tilde{ALPHA}\tilde{CHAIN} PRECURSOR (MHC DN-ALPHA) - HOMO SAPIENS (HUMAN), 250 aa.	Human Gene Homologous to SWISSPROT-ID:P06340 HLA CLASS II HISTOCOMPATIBILITY ANTIGEN, DZ ALPHA CHAIN PRECURSOR (MHC DN-ALPHA) - HOMO SAPIENS (HUMAN), 250 aa.	Human Gene Homologous to SPTREMBL-ID:Q95368 HLA CLASS I INHIBITORY NK RECEPTOR - HOMO SAPIENS (HUMAN), 455 aa.	Human Gene Homologous to SPTREMBL-ID:Q95368 HLA CLASS I INHIBITORY NK RECEPTOR - HOMO SAPIENS (HUMAN), 455 aa.
МНС	МНС	МНС	МНС	МНС
SILENT: CODING	SILENT: CODING	SILENT. CODING	SILENT- CODING	SILENT- CODING
<u> </u>	Gly	Gly	Pro	ng O
<u>a</u>	Gly	Ŋ.	Pro	Olu
H	<u>.</u> ე	E-	<u>-</u>	∢.
U	U .	υ	∢	ຍ
TGGTCTGTGCCCTG GGCCTGGCCAT[C/T JGGCCTGGTGGGCT TCCTCGTGGGCA	TGGGCCTGGCCAT CGGCCTGGTGGG[C /GJTTCCTCGTGGGC ACCGTCCTCATCA	TCGGCCTGGTGGG CTTCCTCGTGGG[C/ TJACCGTCCTCATC ATCATGGGCACAT	GTTTCCTCATTAGC CCTGTGACCCC[AT]GCACACGCAGGG ACCTACAGATGTC	TTGACATCTACCAT CTATCCAGGGA[G/ A]GGGGAAGCCCA TGAACTTAGGCTC C
881	893	905	279	492
cg42686658	cg42686658			cg38337333
08	18	83	83	84

19	61	61	20	3 (3p24)
1.80E-113	1.80E-113	1.80E-113	0	0
Human Gene Homologous to SPTREMBL-ID:Q95368 HLA CLASS I INHIBITORY NK RECEPTOR - HOMO SAPIENS (HUMAN), 455 aa.	Human Gene Homologous to SPTREMBL-ID:Q95368 HLA CLASS I INHIBITORY NK RECEPTOR - HOMO SAPIENS (HUMAN), 455 aa.	Human Gene Homologous to SPTREMBL-ID:Q95368 HLA CLASS I INHIBITORY NK RECEPTOR - HOMO SAPIENS (HUMAN), 455 aa.	Human Gene SPTREMBL- ID:Q14193 H-DRK1 K(+) CHANNEL - HOMO SAPIENS (HUMAN), 858 aa.	Human Gene SPTREMBL- ID:Q14524 SODIUM CHANNEL ALPHA SUBUNIT - HOMO SAPIENS (HUMAN), 2016 aa.
MHC	МНС	МНС	misc_channel	misc_channel
SILENT- CODING	SILENT- CODING	SILENT- CODING	SILENT- CODING	SILENT. CODING
Thr	Phe	Leu	ᄺ	Cys
Th.	Phe	Leu	TT.	Cys
<u> </u>	H	υ	£-	H
T	ပ	1	ວ	U
CTTCTAGTAGTTGG CCTTCACCCAC[T/A)GAACCAAGCTTCA AAACTGGTATCG	GGTACTCAGTGGC CATCATCCTTIC/ TJACCATCCTTCCC TTCTTTCTCCTTC	TGGCCATCCTC TTCACCATCCT[T/C JCCTTCTTTCTCT TCATCGCTGGT	AGGAGCTCAAGCG TGAGGCCGAGACJC TJCTACGGGAGCG GGAAGGCGAGGAG T	TCATGGGCAACCT AAGGCACAGTG CTJGTGCGCAACTT CACAGCGCTCAAC G
669	774	783	649	066
cg38337333			6	cg39660131
\$	86	28		68

19 (19413.1)			(8p11.2)
2,20E-113	7.90E-79	7.90E-79	6.10E-70
Human Gene Homologous to SWISSPROT-ID:Q07699 SODIUM CHANNEL BETA-1 SUBUNIT PRECURSOR - HOMO SAPIENS (HUMAN), 218 aa.lpcls:TREMBLNEW- ID:G2804300 VOLTAGE-GATED SODIUM CHANNEL BETA-1 SUBUNIT - HOMO SAPIENS (HUMAN), 218 aa.	Human Gene Similar to SPTREMBL-ID:P91197 SIMILAR TO LIGAND-GATED IONIC CHANNEL PROTEIN - CAENORHABDITIS ELEGANS, 461 aa.	Human Gene Similar to SPTREMBL-ID:P91197 SIMILAR TO LIGAND-GATED IONIC CHANNEL PROTEIN - CAENORHABDITIS ELEGANS, 461 aa.	Human Gene Similar to SPTREMBL-ID:P91197 SIMILAR TO LIGAND-GATED IONIC CHANNEL PROTEIN - CAENORHABDITIS ELEGANS, 461 aa.
misc_channel	misc_channel	misc_channel	misc_channel
SILENT. CODING	SILENT- CODING	SILENT- CODING	SILENT- CODING
<u>n</u> 10	Asp	Çys	Pro
Glu	Asp	ζ _ζ	Pro
O	Į	H	O .
V	U	U	∀
CGGAATACCTGGC CATCACCTCTGA[A/ GJAGCAAAGAGAA CTGCACGGGCGTC C	AGAGTGGCGAGTG GGTCATCGTGGA[C /TJGCCGTGGGCAC CTACAACACCAGG A	ACAACACCAGGAA GTACGAGTGCTG[C /T]GCCGAGATCTA CCCGGACATCACC T	AGAGGCTCTTTCTG CAGAAACTTCC[A CJAAATTACTTTGC ATGAAAGATCATG
717	870	606	1160
cg44963814	cg21413267	cg21413267	og3000465
	91	92	93

11 (11922)	(11q22)		14 (14q24)
	0	1.10E-115	0
Human Gene SWISSNEW- ID:P06401 PROGESTERONE RECEPTOR (PR) - HOMO SAPIENS (HUMAN), 933 aa.lpcls:SWISSPROT-ID:P06401 PROGESTERONE RECEPTOR (PR) - HOMO SAPIENS (HUMAN), 933 aa.	Human Gene SWISSNEW- ID:P06401 PROGESTERONE RECEPTOR (PR) - HOMO SAPIENS (HUMAN), 933 aa.lpcls:SWISSPROT-ID:P06401 PROGESTERONE RECEPTOR (PR) - HOMO SAPIENS (HUMAN), 933 aa.	Human Gene Homologous to SWISSPROT-ID:P07992 DNA EXCISION REPAIR PROTEIN ERCC-1 - HOMO SAPIENS (HUMAN), 297 aa.	Human Gene SPTREMBL- ID:Q99907 LATENT TRANSFORMING GROWTH FACTOR-BETA-BINDING PROTEIN-2 - HOMO SAPIENS (HUMAN), 1821 aa.
nucl_recpt	nucl_recpt	nuclease	oncogene
SILENT- CODING	SILENT- CODING	SILENT. CODING	SILENT- CODING
His	Gin	Asn	Thr.
His	ul D	Asn	Th
[<u> </u>	Ö	<u> </u>
v	¥	⋖	∢
GTCTAGGATGGAG ATCCTACAACA[C TJGTCAGTGGGCA GATGCTGTATTTTG	ATAACTTGCATGA TCTTGTCAAACA[A/ G]CTTCATCTGTAC TGCTTGAATACAT	TTACGTCGCCAAA TTCCCAGGGCACIA /GJTTGCGCACGAA CTTCAGTACGGGA T	TCCCTGTGACCCA GGCAGGTGCATG[A /G]GTGACACTGGT CGTGACCTGGCCA G
3766	4114	713	4226
	og30421838	cg43947341	cg43939230
94	95	96	97

	22 (22q11)	(1925)	
3.70E-182	2.40E-84	2.80E-287	2.40E-155
Human Gene SWISSPROT- ID:P31314 HOMEOBOX PROTEIN HOX-11 (TCL-3 PROTO-ONCOGENE) - HOMO SAPIENS (HUMAN), 330 aa.	Human Gene Similar to SWISSPROT-ID:Q64010 PROTO- ONCOGENE C-CRK (P38) (ADAPTER MOLECULE CRK) - MUS MUSCULUS (MOUSE), 304 aa.	Human Gene SWISSPROT- ID: P19878 NEUTROPHIL CYTOSOL FACTOR 2 (NCF-2) (NEUTROPHIL NADPH OXIDASE FACTOR 2) (P67- PHOX) - HOMO SAPIENS (HUMAN), 526 aa.	Human Gene SWISSPROT- ID:P00491 PURINE NUCLEOSIDE PHOSPHORYLASE (EC 2.4.2.1) (INOSINE PHOSPHORYLASE) (PNP) - HOMO SAPIENS (HUMAN), 289 aa.
oncogene	oncogene	oxidase	phosphorylase
SILENT. CODING	SILENT: CODING	SILENT: CODING	SILENT- CODING
Ala	<u>≅</u>	Leu	Pro
Ala	e = = = = = = = = = = = = = = = = = = =	Leu	Pro
£-s	H	F	₹
Ö	U	ပ	ර
CGGCACACAGGCC IC GCTCGCCGGAGCIC /TJGTGGCCCACCCC CAGCCCTGGCCA	AGAACTCGCGGGT CTCCCACTACAT[C/ TJATCAACTCGCTG CCCAACCGCCGTT	CTGCAACTACCTTG AACCAGTTGAG[C/ TJTGCGGATCCACC CTCAGCAGCAGCC	CAGCATGACCTGG CACTGTACTTCG[G/ AJGGAAAGTTGGG GATTTCACCGTAGT
1447	742	963	1310
cg42674136	cg41972699	cg42849556	cg43996195
86	66	100	101

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	lo.
2.40E-155	5.40E-62
Human Gene SWISSPROT- ID:P00491 PURINE NUCLEOSIDE PHÓSPHORYLASE (EC 24.2.1) (INOSINE PHOSPHORYLASE) (FNP) - HOMO SAPIENS (HUMAN), 289 aa.	Human Gene Similar to SWISSNEW-ID:P53999 ACTIVATED RNA POL YMERASE II TRANSCRIPTIONAL COACTIVATOR PI S (PC4) (P14) - HOMO SAPIENS (HUMAN), 126 aa, pcls:SWISSPROT- ID:P53999 ACTIVATED RNA POL YMERASE II TRANSCRIPTIONAL COACTIVATOR PI S (PC4) (P14) - HOMO SAPIENS (HUMAN), 126 aa.
SILENT- phosphorylase CODING	polymerase
SILENT- CODING	SILENT- CODING
His	DI.
His]E
₹	· ·
Ø	V
TTGCAACTTGAGG G TCGGTGCTTAGT[G/ AJTGAGACAGAAG CCATTCTGCAGTGT	TTTACAGTTTTCTT ACTGCATCATC[AT JATGTCAGAAATCT GTTCCTTCAGCT
,	372
cg43996195 1421	cg43948227
102	103

,
4.405-241
potassium_chan Human Gene SWISSNEW- ID:P48050 INWARD RECTIFIER POTASSIUM CHANNEL 4 (POTASSIUM CHANNEL, INWARDL Y RECTIFYING, SUBFAMIL Y I, MEMBER 4) (HIPPOCAMPAL INWARD RECTIFIER) (HIR) (HRK1) (HIRK2) (KIR2.3) - HOMO SAPIENS (HUMAN), 445 aa_lpcis.SWISSPROT-ID:P48050 INWARD RECTIFIER POTASSIUM CHANNEL 4 (POTASSIUM CHANNEL, INWARDLY RECTIFYING, SUBFAMILY I, MEMBER 4) (HIRK2) (KIR2.3) - HOMO SAPIENS (HUMAN), 445 aa.
potassium_chan
CODING
ಕ್ರ ഗ
es N
0
AGAGCCACTACAA A GGTGGACTACTCIA AGCTACGAGGTGG ACCTACGAGGTGG
cg43333426 1302
104

2	14 (14q32.1)
1.20E-208	4.40E-83
Human Gene SWISSPROT- ID:P07093 GLIA DERIVED NEXIN PRECURSOR (GDN) (PROTEASE NEXIN I) (PN-1) (PROTEASE INHIBITOR 7)- HOMO SAPIENS (HUMAN), 398 aa.	Human Gene Similar to SWISSPROT-ID:P17475 ALPHA- 1-ANTIPROTEINASE PRECURSOR (ALPHA-1- ANTITRYPSIN) (ALPHA-1- PROTEINASE INHIBITOR) - RATTUS NORVEGICUS (RAT), 411 aa.
proteaseinhib	proteaseinhib
SILENT- CODING	SILENT- CODING
Val	Thr
Val	Thr
ტ	[
υ	ت د
GCAGGATCACCTG CACCCTCTTGGG[C/ GJACCATGATGCTC ATCCAGCTGTCTA	AGTCAGACACCAG CTTAGAAATGAC[C /T]ATGGGCAATGC CTTGTTTCTTGATG
1081	624
cg43920929	cg43059041
106	107
	cg43920929 1081 GCAGGATCACCTG C G Val Val SILENT- proteaseinhib Human Gene SWISSPROT- 1.20E-208 CACCCTCTTGGG[C/ CACCCTCTTGGG[C/ CODING D.P07093 GLIA DERIVED NEXIN PRECURSOR (GDN) GJACCATGATGCTC ATCCAGCTGTCTA (PROTEASE NEXIN I) (PN-1) (PROTEASE NHIBITOR 7)- HOMO SAPIENS (HUMAN), 398 Aa.

3 (3p)	17	7	20 (20q12)	œ
2.90E-260	1.40E-180	2.40E-114	1.50E-80	8.30E-58
Human Gene SPTREMBL- ID:Q92777 SYNAPSIN IIB - HOMO SAPIENS (HUMAN), 478 aa.	Human Gene SPTREMBL- ID:Q28686 50-KDA DYSTROPHIN-ASSOCIATED GLYCOPROTEIN PRECURSOR - ORYCTOLAGUS CUNICULUS (RABBIT), 387 aa.	Human Gene Homologous to TREMBLNEW-ID: G1703715 PANTOPHYSIN-SYNAPTOPHY SIN HOMOLOG - MUS SP, 261 aa.	Human Gene Similar to SWISSPROT-ID:P02585 TROPONIN C, SKELETAL MUSCLE - HOMO SAPIENS (HUMAN), 159 aa.	Human Gene Similar to SWISSPROT-ID:P02535 KERATIN, TYPE I CYTOSKELETAL 10 (CYTOKERATIN 10) (56 KD (CYTOKERATIN 10) (56 KD TYPE I CYTOSKELETAL 59 KD) - MUS MUSCULUS (MOUSE), 569 aa.
struct	struct	struct	struct	struct
SILENT- CODING	SILENT- CODING	SILENT- CODING	SILENT- CODING	SILENT- CODING
Leu	njo J	Leu	<u>1</u>	Arg
Leu	Olu	Leu	Th	Arg
T	∢	_ව	∢	ى ت
ပ	ප	∢	ပ ပ	O
GGAGGACAGGCAA CTCATCACCGAA[C M]TAGTCATCAGCA AGATGAACCAGCT	ACCCGTTCTTCTGC CCACCCACTGA[G/ A]GCCCCAGACCGT GACTTCTTGGTGG	TCTGGAAGCCGGA CATCCTCTGAGC[A/ GJAGTCGACTGATC CGCTGGCGAACCA	TCATCAGAGATTC GATCTCCTCGTC[C/ A]GTCACGTGCTCC CCGGAGGCCCTGA	GCTTTGAGGAGGA GGCGCGGTTGCG[C /G]GACGACACTGA GGCGGCCATCCGC G
1385	7001	2160	497	788
cg40148056	cg42894986	cg43961212	 	cg43960684
108	109	110	111	112

∞	16
9.20E-83	7.70E-79
Human Gene Similar to SPTREMBL-ID:Q42761 SQUALENE SYNTHASE (EC 2.5.1.21) (FARNESYL- DIPHOSPHATE FARNESYLTRANSFERASE) (FARNESYLTRANSFERASE) (FRREQUALENE-DI- DIPHOSPHOSPHATE SYNTHASE) - GLYCYRRHIZA GLABRA, 412 aa.	Human Gene Similar to SWISSNEW-ID:P39062 ACETYL-COENZYME A SYNTHETASE (EC 6.2.1.1) (ACETATE-COA LIGASE) (ACYL- ACTIVATING ENZYME) (ACETYL-COA SYNTHASE) - BACILLUS SUBTILIS, 572 aa.pcis:SWISSPROT-ID:P39062 ACETYL-COENZYME A SYNTHETASE (EC 6.2.1.1) (ACETATE-COA LIGASE) (ACYL- ACTIVATING ENZYME) (ACETYL-COA SYNTHASE) - BACILLUS SUBTILIS, 572 aa.
synthase	synthase
SILENT- CODING	SILENT- CODING
Leu	Ď
Leu	Ġ
o	E-
O	·
TTCGGAAAGGGCA G AGCAGTGACCCT[6 /C]ATGATGGATGC CACCAATATGCCA G	ACACCCACAGCAG TTTTGGTTTAGG[A/ TJTTATCTGTAAAT GGAAGGTTCTGGC
1049	901
cg43958714	cg43124627
113	114

9.908-70
Human Gene Similar to SWISSNEW-ID:P53556 8- AMINO-7-OXONONANOATE SYNTHASE (EC 2.3.1.47) (7- KETO-8-AMINO-PELARGONIC ACID SYNTHETASE) (7-KAP SYNTHETASE) (L-ALANINE- PIMELYL COA LIGASE) - BACILLUS SUBTILIS, 389 aa.pcis:SWISSPROT-ID:P53556 8-AMINO-7-OXONONANOATE SYNTHASE (EC 2.3.1.47) (7- KETO-8-AMINO-PELARGONIC ACID SYNTHETASE) (L-ALANINE- PIMELYL COA LIGASE) - BACILLUS SUBTILIS, 389 aa.
synthase
SILENT - synthase CODING
V I I I I I I I I I I I I I I I I I I I
Ala
←
V
TCTTCTCCAACAGT A CTGCCACCGC[A/T JGTCGTTGGCTGCG CCTCCAAGGCCC
15 cg43968419 906
115

7.40E-65	7.40E-65
Human Gene Similar to SWISSNEW-ID:P39062 ACETYL-COENZYME A SYNTHETASE (EC 6.2.1.1) (ACETATE—COA LIGASE) (ACYL- ACTIVATING ENZYME) (ACETYL-COA SYNTHASE) - BACILLUS SUBTILIS, 572 aa, pecis:SWISSPROT-ID:P39062 ACETYL-COENZYME A SYNTHETASE (EC 6.2.1.1) (ACETATE—COA LIGASE) (ACYL- ACTIVATING ENZYME) (ACETYL-COA SYNTHASE) - BACILLUS SYNTHASE) - BACILLUS SYNTHASE) - BACILLUS	Human Gene Similar to SWISSNEW-ID:P39062 ACETYL-COENZYME A SYNTHETASE (EC 6.2.1.1) (ACETATE-COA LIGASE) (ACYL- ACTIVATING ENZYME) (ACETYL-COA SYNTHASE) - BACILLUS SUBTILIS, 572 aa. pcls:SWISSPROT-ID:P39062 ACETYL-COENZYME A SYNTHETASE (EC 6.2.1.1) (ACETATE-COA LIGASE) (ACYL- ACTIVATING ENZYME) (ACETYL-COA SYNTHASE) - BACILLUS SUBTILIS, 572 aa.
synthase	synthase
SILENT- CODING	CODING
Leu	Lys
Leu	Lys
≺	V
<u>o</u>	O
6) 4	TCACAGGAAAAT (TCAACGAGCCA[G /A]CTTCGAGACAA GGAGTGGAAGATG T
1484	
	cg43064068
116	117

==	=	=	4	4
1.70E-241	1.70E-241	1.70E-241	1.60E-236	1.60E-236
Human Gene SWISSPROT- ID:P14416 D(2) DOPAMINE RECEPTOR - HOMO SAPIENS (HUMAN), 443 aa.	Human Gene SWISSPROT- ID:P14416 D(2) DOPAMINE RECEPTOR - HOMO SAPIENS (HUMAN), 443 aa.	Human Gene SWISSPROT- ID:P14416 D(2) DOPAMINE RECEPTOR - HOMO SAPIENS (HUMAN), 443 aa.	Human Gene SWISSPROT- ID:P25101 ENDOTHELIN-1 RECEPTOR PRECURSOR (ET-A) - HOMO SAPIENS (HUMAN), 427 aa.	Human Gene SWISSPROT- ID:P25101 ENDOTHELIN-1 RECEPTOR PRECURSOR (ET-A) - HOMO SAPIENS (HUMAN), 427 aa.
tm7	m7.	Tan	TEL 1	tm.7
SILENT- CODING	SILENT- CODING	SILENT- CODING	SILENT- CODING	SILENT- CODING
H H	nen Ten	Pro	H	Glu
His	Leu	Pro	His	Glu
H	F-	∢	<u> </u>	ن ن
ပ	U	o ·	υ	∢
TGACTCTCCCCGAC CCGTCCCACCA[CT] JGGTCTCCACAGCA CTCCCGACAGCC	TCCGCAAGGCCTT CCTGAAGATCCT[C/ T]CACTGCTGACTC TGCTGCCTGCCCG	TCCTCGTCGCCACA CTGGTCATGCC[C/A JTGGGTTGTCTACC TGGAGGTGGTAG	TTGCTCTTTGCTGG TTCCCTCTTCA[CT] TTAAGCCGTATATT GAAGAAAACTG	TATTGAAGAAAAC TGTGTATAACGA[A /G]ATGGACAAGAA CCGATGTGAATTA C
1278	1662	909	1471	1507
cg41084924	cg41084924	cg41084924	cg43985000	cg43985000
118		120	121	122

	
	*
5.00E-217	3.00E-212
Human Gene SWISSPROT- ID:P3089 NEUROTENSIN RECEPTOR TYPE I (NT-R-1) (HIGH-AFFINITY LEVOCABASTINE- INSENSITIVE NEUROTENSIN RECEPTOR) (NTRH) - HOMO SAPIENS (HUMAN), 418 aa.	Human Gené SWISSNEW- ID:P32247 BOMBESIN RECEPTOR SUBTYPE-3 (BRS-3) - HOMO SAPIENS (HUMAN), 399 aa_[pcls:SWISSPROT- ID:P32247 BOMBESIN RECEPTOR SUBTYPE-3 (BRS-3) - HOMO SAPIENS (HUMAN), 399 aa_[pcls:TREMBLNEW- ID:E1240254 BOMBESIN RECEPTOR SUBTYPE-3 (UTERNINE BOMBESIN RECEPTOR, SUBTYPE-3 (UTERNINE BOMBESIN RECEPTOR, SUBTYPE-3 (UTERNINE BOMBESIN RECEPTOR, BRS-3) - HOMO SAPIENS (HUMAN), 399 aa.
fm7	tm7
SILENT- CODING	SILENT- CODING
Lys	Tyr
Lys	Tyr
∢ .	· [
O	U
ACGTGAACACCGA G CATCTACTCCAA[G/ A]GTGCTGGTGACC GCCGTGTACCTGG	ATTCCTTGATTGCT AGGACCCTTTA[CT JAAAAGCACCCTGA ACATACCTACTG
561	1263
cg44930578 561	cg3003519
	124

×	12 (14q32.1)	19 (19q13.3)	19 (19q13.3)
3.005-212	9.00E-211	8.30E-208	8.30E-208
Human Gene SWISSNEW- ID:P32247 BOMBESIN RECEPTOR SUBTYPE-3 (BRS-3) - HOMO SAPIENS (HUMAN), 399 aa,pcis:SWISSPROT- ID:P32247 BOMBESIN RECEPTOR SUBTYPE-3 (BRS-3) - HOMO SAPIENS (HUMAN), 399 aa,pcis:TREMBLNEW- ID:E1240254 BOMBESIN RECEPTOR SUBTYPE-3 (UTERINE BOMBESIN RECEPTOR SUBTYPE-3 (UTERINE BOMBESIN RECEPTOR (HUMAN), 399 aa.	Human Gene SWISSPROT- ID:P30411 B2 BRADYKININ RECEPTOR (BK-2 RECEPTOR) - HOMO SAPIENS (HUMAN), 391 aa.	Human Gene SWISSPROT- ID:P43119 PROSTACYCLIN RECEPTOR (PROSTANOID IP RECEPTOR) (PGI RECEPTOR) - HOMO SAPIENS (HUMAN), 386 aa.	Human Gene SWISSPROT- ID:P43119 PROSTACYCLIN RECEPTOR (PROSTANOID IP RECEPTOR) (PGI RECEPTOR) - HOMO SAPIENS (HUMAN), 386 aa.
tm7	tm.7		m7
CODING CODING	SILENT: CODING	SILENT- CODING	SILENT. CODING
Ą	Tyr	Sci	Val
Gjy.	Tyr	Ser	Val
E	<u></u>	∀	o
O	ပ	O	U
CTTATGCTGTGATC ATTTCAGTGGG[C/T JATCCTTGGAAATG CTATTCTCATCA CTATTCTCATCA	TCCGAAAGAAGTC TTGGGAGGTGTA[C /T]CAGGGAGTGTG CCAGAAAGGGGGC T	AGACACCCTTTCC CAGCTCGCTA JGGGAGGAGGGAC CCAAGGGCCCCCT	GCCCTCGGCCTTC GCGGTGCTGGT[C/ G]ACCGGACTGGCG GCCACCGACCTGC
711	1182	1097	272
	cg43969010	cg43263108	cg43263108
125	126	127	128

8 (8q11.2)	8 (8q11.2)	8 (8q11.2)		1 (1p36.1)
2.10E-204	2.10E-204	2.10E-204	1.405-196	2.10E-195
Human Gene SWISSPROT- ID:P41145 KAPPA-TYPE OPIOID RECEPTOR (KOR-1) - HOMO SAPIENS (HUMAN), 380 aa.	Human Gene SWISSPROT- ID:P41145 KAPPA-TYPE OPIOID RECEPTOR (KOR-1) - HOMO SAPIENS (HUMAN), 380 aa.	Human Gene SWISSPROT- ID:P41145 KAPPA-TYPE OPIOID RECEPTOR (KOR-1) - HOMO SAPIENS (HUMAN), 380 aa.	Human Gene TREMBLNEW- ID:G2736282 G PROTEIN COUPLED RECEPTOR - HOMO SAPIENS (HUMAN), 362 aa.	Human Gene SWISSPROT- ID:P41143 DELTA-TYPE OPIOID RECEPTOR (DOR-1) - HOMO SAPIENS (HUMAN), 372 aa.
rm7	tm7	tm7	tm7	tm7
SILENT- CODING	SILENT- CODING	SILENT- CODING	SILENT: CODING	SILENT. CODING
Ala	Ile	Pro	Ser	Giy
Ala	Ile	Pro	Ser	Gly
O	E	<u>E</u>	ပ	L
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CCAGACTGGTCCT GGTGGTGGTGGCJA /GJGTCTTCGTCGTC TGCTGGACTCCCA	CAGCACTCACCAT GGAATCCCCGATC AJCAGATCTTCCGC GGGGAGCCGGGCC	CGATCCAGATCTTC CGCGGGGAGCC[G/ TJGGCCCTACCTGC GCCCGAGCGCCT	TGGATCTGCACCTC TTCGACTACTC[A/C JGAGCCAGGGAAC TTCTCGGACATCA	CGCTGCACCTGTG CATCGCGCTGGG[C TITACGCCAATAG CAGCCTCAACCC G
1220	392	413	155	1154
cg43267238	cg43267238	cg43267238	cg43264978	cg3001696
129	130	131	132	133

1 (1p36.1)	1 (1p31.2)	-		
2.10E-195	3.10E-194	1.10E-173	2.50E-160	2.50E-160
Human Gene SWISSPROT- ID:P41143 DELTA-TYPE OPIOID RECEPTOR (DOR-1) - HOMO SAPIENS (HUMAN), 372 aa.	Human Gene SWISSPROT- ID:P43115 PROSTAGLANDIN E2 RECEPTOR, EP3 SUBTYPE (PROSTANOID EP3 RECEPTOR) (PGE RECEPTOR, EP3 SUBTYPE) - HOMO SAPIENS (HUMAN), 390 aa.	Human Gene \$WISSPROT- ID:P30542 ADENOSINE A1 RECEPTOR - HOMO SAPIENS (HUMAN), 326 aa.	Human Gene TREMBLNEW- ID:E1246031 OLFACTORY RECEPTOR - HOMO SAPIENS (HUMAN), 312 aa.	Human Gene TREMBLNEW- 1D:E1246031 OLFACTORY RECEPTOR - HOMO SAPIENS (HUMAN), 312 aa.
tm7	tm7	tm7	tm7	m7
SILENT- CODING	SILENT- CODING	SILENT- CODING	SILENT- CODING	SILENT. CODING
Gly	Leu	Val	Thr.	Gly
Gly	3	Val	Thr	Gly
L	ڻ ت	<u>ن</u>	<u>o</u>	U
O	U	U	4	L
TGGCTGTGACCCG TCCCCGGGACGG[G //JGCAGTGGTGTG CATGCTCCAGTTCC	TGGCCTTCCCGATC ACCATGCTGCT[C/G JACTGGTTTCGTGG GCAACGCACTGG	GGGATGCCACCTT CTGCTTCATCGT[C/ G]TCGCTGGCGGTG GCTGATGTGGCCG	CCATCTCCTTCTGT GGCTGTCTCAC[A/ GJCAGATGTATTTC GTTTTCATGTTCG	GGTGGAAAGCCTT CTCCACCTGTGG[T/ CJTCTCACCTGGCT GTGGTTCTCCTCT
815	407	347	358	787
cg3001696	cg42704646	cg43326635	cg3003708	cg3003708
134	135	136	137	138

2.50E-160	1.90E-153	2.10E-67	2.10E-67	2.10E-67
Human Gene TREMBLNEW- ID:E1246031 OLFACTORY RECEPTOR - HOMO SAPIENS (HUMAN), 312 aa.	Human Gene SWISSPROT- ID:Q15062 OLFACTORY RECEPTOR-LIKE PROTEIN FAT11 - HOMO SAPIENS (HUMAN), 316 aa.	Human Gene Similar to SWISSPROT-ID:P30975 TACHYKININ-LIKE PEPTIDES RECEPTOR 99D (DTKR) - DROSOPHILA MELANOGASTER (FRUIT FLY), 519 aa.	Human Gene Similar to SWISSPROT-ID:P30975 TACHYKININ-LIKE PEPTIDES RECEPTOR 99D (DTKR) - DROSOPHILA MELANOGASTER (FRUIT FLY), 519 aa.	Human Gene Similar to SWISSPROT-ID:P30975 TACHYKININ-LIKE PEPTIDES RECEPTOR 99D (DTKR) - DROSOPHILA MELANOGASTER (FRUIT FLY), 519 aa.
tm7	tm7	tm7	tm <i>7</i>	tm7
SILENT- CODING	SILENT- CODING	SILENT: CODING	SILENT. CODING	SILENT- CODING
Phe	Ser	Leu .	His	Gin
Phe	Ser	Leu .	His	Gln
0	H	ල ·	<u>o</u>	4
H	ပ	ပ	£	ຽ
ACAGCACCATCAT TGCTGTGTATTITI CJAACCCTCTGTCC TCCCACTCAGCTG	ACTCTCCAATGTAG TTTTTCCTCTC[C/T] AACCTCTCCTTCTT GGACCTCTGCT	GACATCAGGCGCA CGGTGCCAACCT[C /G]CGCCATCTGCA GGCCAAGAAGAAG T	AGGCGCACGGTGC CAACCTCCGCCAII/ CJCTGCAGGCCAAG AAGAAGTTTGTGA	CAGCCTTCTCCATG CCCAGCTGGCA[G/ A]CTGGCACTGTGG GCACCAGCCTACC
841	537	717	723	96
c <u>g</u> 3003708				cg38841806
139	140	141	142	143

<i>5</i> (5q32)	5 (5q32)	3 (3925)	15
2.00E-58	2.00E-58	2.20E-207	9.00E-179
Human Gene Similar to SWISSPROT-ID:Q24563 DOPAMINE RECEPTOR 2 - DROSOPHILA MELANOGASTER (FRUIT FLY), 539 aa.	Human Gene Similar to SWISSPROT-ID:Q24563 DOPAMINE RECEPTOR 2 - DROSOPHILA MELANOGASTER (FRUIT FLY), 539 aa.	Human Gene SWISSPROT- ID:P26022 PENTAXIN- RELATED PROTEIN PTX3 PRECURSOR (TUMOR NECROSIS FACTOR- INDUCIBLE PROTEIN TSG-14) - HOMO SAPIENS (HUMAN), 381	Human Gene SWISSPROT- ID:Q06545 GA BINDING PROTEIN BETA-2 CHAIN (GABP-BETA-2 SUBUNIT) (TRANSCRIPTION FACTOR E4TF1-47) (GAPBP2) - HOMO SAPIENS (HUMAN), 347 aa.
m7	m7	ju	transcriptfactor
SILENT- CODING	SILENT- CODING	SILENT- CODING	SILENT: CODING
Leu	Arg	Pro	Lys
Leu	Arg	Pro	Lys
⋖	<u> </u>	-	Ö
U	ပ	⋖	∢
CCTGTGCTGATCTG G GTCATGGGCCT[G/ A]GCAGTGGTGCCC TTTGGGGCCGCCC	CTTGCCCATTCAGA C TGCACTGGTAC[C/ A]GGGCCACCCACC AGGAAGCCATCAA	TTGGAAGCGTGCA , TCCAGTGAGACC[A T]ATGAGGCTTGA GTCTTTTAGTGCCT	GTGTGAGCAGAGA / TGCCAGAACCAA[A /G]GTGGACCGAAC ACCATTACATATG G
1966	2237	687	376
cg43040273	cg43040273	cg43336100	cg21646034
144	145	146	147

	34)
~	9 (9434)
5.30E-245	6.50E-192
Human Gene SWISSPROT- ID-1939656 DOLICHYL- DIPHOSPHOOLIGOSACCHARI DE-PROTEIN GLYCOSYLTRANSFERASE 48 KD SUBUNIT PRECURSOR (EC 2.4.1.119) (OLIGOSACCHARYL TRANSFERASE 48 KD SUBUNIT) (DOOST 48 KD SUBUNIT) (KIAA0115) (HA0643) - HOMO SAPIENS (HUMAN), 456 aa.	Human Gene SWISSPROT- D:P16442 FUCOSYLGLYCOPROTEIN ALPHA-N- ALPHA-N- ACETYLGALACTOSAMINYLT RANSFERASE (BC 2.4.1.40) (HISTO-BLOOD GROUP A TRANSFERASE) (A TRANSFERASE) / FUCOSYLGLYCOPROTEIN 3- ALPHA- GALACTOSYLTRANSFERASE (BC 2.4.1.37) (HISTO-BLOOD GROUP B TRANSFERASE) (B
transferase	transferase
SILENT	SILENT. CODING
Gly	년 된
Gly	Thr
g	O
V	V
TGGCAGCTACCAG CACACTGCTCC[A/ GJCCGTCAATAAAG GCACTGATGGTCT	TGGCTCCCATTGTC TGGGAGGCCACIA/ GJTTCAACATCGAC ATCCTCAACGAGC
1608	294
2	cg2537639
148	149

.9 (9q34)
6.50E-192 [.9 (9q34)
Human Gene \$WISSPROT- D.P16442 FUCOSYLGLYCOPROTEIN ALPHA-N- ACETYLGALACTOSAMINYLT RANSFERASE (EC 2.4.1.40) (HISTO-BLOOD GROUP A TRANSFERASE) (A TRANSFERASE) (A TRANSFERASE) (A TRANSFERASE) (A TRANSFERASE) (A TRANSFERASE) (B TRANSFERASE) (B TRANSFERASE) GALACTOSYLTRANSFERASE (EC 2.4.1.37) (HISTO-BLOOD GROUP B TRANSFERASE) (B TRANSFER
transferase
SILENT- CODING
His
His
H
O
ACGTGGACATGGA C GTTCCGCGACCA[C //JGTGGGCGTGGA GATCCTGACTCCG C
654
cg2537639

9 (9q34)	
6.50E-192	
Human Gene SWISSPROT- ID:P16442 FUCOSYLGLYCOPROTEIN ALPHA-N- ALPHA-N- ACETYLGALACTOSAMINYLT RANSFERASE (EC.2.4.1.40) (HISTO-BLOOD GROUP A TRANSFERASE) (A TRANSFERASE) / FUCOSYLGLYCOPROTEIN 3- ALPHA- GALACTOSYLTRANSFERASE (GC.2.4.1.37) (HISTO-BLOOD GROUP B TRANSFERASE) (B TRANSFERASE) (MAGAT) - HOMO SAPIENS (HUMAN), 354 aa.	
SILENT- transferase CODING	
SILENT- CODING	
7 0	:
Pro	- ,
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U	
ACGTGGGCGTGGA GATCCTGACTCGG/ AJCTGTTCGGCACC CTGCACCCCGGCT	
•	
cg2537639 678	
151	·

9 (9q34)										_				-							
6.50E-192 9 (9q34)									_												
Human Gene SWISSPROT-	ID:P16442	FUCOSYLGLYCOPROTEIN	ALPHA-N-	ACETYLGALACTOSAMINYLT	RANSFERASE (EC 2.4.1.40)	(HISTO-BLOOD GROUP A	TRANSFERASE) (A	TRANSFERASE)/	FUCOSYLGLYCOPROTEIN 3-	ALPHA-	GALACTOSYLTRANSFERASE	(EC 2.4.1.37) (HISTO-BLOOD	GROUP B TRANSFERASE) (B	TRANSFERASE) (NAGAT) -	HOMO SAPIENS (HUMAN), 354	aa.					
transferase																					
SILENT- transferase	CODING					ı												,			
Pro																					
Pro																		,			
T					-					_							<u>.</u>		 	_	
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GGCCCCAGTCCCA	GGCCTACATCCC[C/	TJAAGGACGAGGG	CGATTTCTACTACC																		
l																					
cg2537639 768)				-											-					
152																					

9 (9q34)	9
6.50E-192	1.60E-117
Human Gene SWISSPROT- D:P16442 FUCOSYLGL YCOPROTEIN ALPHA-N- ACETYLGALACTOSAMINYLT RANSFERASE (EC 2.4.1.40) (HISTO-BLOOD GROUP A TRANSFERASE) (A TRANSFERASE) (A TRANSFERASE) (A TRANSFERASE) (A TRANSFERASE) (A TRANSFERASE) (A TRANSFERASE) (B TRANSFERASE) FUCOSYLGL YCOPROTEIN 3- ALPHA- GALACTOSYL TRANSFERASE (EC 2.4.1.37) (HISTO-BLOOD GROUP B TRANSFERASE) (B TRANSFE	Human Gene Homologous to SWISSPROT-ID:P30711 GLUTATHIONE S- TRANSFERASE THETA 1 (EC 2.5.1.18) (CLASS-THETA) - HOMO SAPIENS (HUMAN), 239 aa.
transferase	transferase
SILENT- CODING	SILENT- CODING
F. F.	Val
Leu	Val
₹	၁
(D)	V
ACGAGAGCCACCT GAACAAGTACCT[G /a]CTGCGCCACAA ACCCACCAAGGTG C	GGGGGGATACTGG CTCACCCAGGAA[A /C]ACAGGGAACAT CACCTTATGCCAC A
927	732
	cg44000740
153	154

	ନ୍ତି		
13	5 (5p15.3)	1	
0	0	1.60E-259	4.40E-241
Human Gene SWISSPROT- ID:P30825 HIGH-AFFINITY CATIONIC AMINO ACID TRANSPORTER-1 (CAT-1) (CAT1) (SYSTEM Y+ BASIC AMINO ACID TRANSPORTER) (ECOTROPIC RETROVIRAL LEUKEMIA RECEPTOR HOMOLOG) (ERR) (ECOTROPIC RETROVIRUS RECEPTOR HOMOLOG) - HOMO SAPIENS (HUMAN), 629 aa.	Human Gene \$WISSPROT- ID:Q01959 SODIUM- DEPENDENT DOPAMINE TRANSPORTER (DA TRANSPORTER) (DAT) - HOMO SAPIENS (HUMAN), 620 aa.	Human Gene SWISSPROT- ID:P11166 GLUCOSE TRANSPORTER TYPE 1, ERYTHROCYTE/BRAIN - HOMO SAPIENS (HUMAN), 492 aa.	Human Gene SPTREMBL- ID:Q14728 TETRACYCLINE TRANSPORTER-LIKE PROTEIN MRNA - HOMO SAPIENS (HUMAN), 455 aa.
fransport	transport	transport	transport
SILENT- CODING	SILENT.	SILENT. CODING	SILENT. CODING
స	Ser	Asp	Pro
(%)	Ser	Asp	Pro
[m	ල	K .	Ö
		5	
ACGCAGTGGCCGT C GGGCTCCCTCTG[C/ TJGCTCTTTCCGCC AGTCTTCTAGGTT	CCATCGCCACGCT A CCCTCTGTCCTC[A/ G]GCCTGGGCCGTG GTCTTCTTCATCA	GATGGAACAGCTC CTCGGGTGTCTT[G/ AJTCACTTTGGCTG GCTCCCCCTGCC	GCGGCTGCTGGTG GATGGTGGGCG[C /G]GGGGTGCAGCC TCCACCCCTCCCC
1185	1347	1719	1656
cg38869466	cg40351913	cg43964039	cg43992017
155	156	157	158

	9	9	_	2 (2q34)	
	10	16		2	
0	0	0	0	0	
Human Gene SWISSNEW- ACC:Q15031 PROBABLE LEUCYL-TRNA SYNTHETASE, MITOCHONDRIAL PRECURSOR (EC 6.1.1.4) (LEUCINE-TRNA LIGASE) (LEUCINE-TRNA LIGASE) (LEURS) (KIAA0028) - Homo sapiens (Human), 903 aa.	Human Gene SPTREMBL. ACC:Q16084 P130 - HOMO SAPIENS (HUMAN), 1139 aa.		Human Gene SWISSPROT- ACC:P42694 HYPOTHETICAL PROTEIN KIAA0054 - Homo sapiens (Human), 1942 aa.	Human Gene REMTREMBL- ACC:E1296438 SEQUENCE 28 FROM PATENT WO9727323 - UNIDENTIFIED, 1829 aa.	
UNCLASSIFIE D	UNCLASSIFIE D	UNCLASSIFIE D	UNCLASSIFIE D	UNCLASSIFIE D	
SILENT- CODING	SILENT- CODING		SILENT- CODING	SILENT- CODING	
<u>s</u>	Arg	Ala	Gly	Ala	
Leu	Arg	Ala	Gly	Ala	
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CGCCTGTAATGGC TGTGAACATGCT[C/ TJACCCAGCAGGAG GTCCCTGTCGTTA	CATTGACTAGGG CTGTGGGGGCAT[C /G]CGCCCAGGTGT CCCTCCATCAGAG G	AGCAGGCCAAGAG AGATCTGTGGAA[C /TJGCATCTTGTTCC AGAATACCAGATA	CGCTGGCATAGGA CATGGCGGGCTT[G ATCCCCCCGCAGA GCTCTGGGGGGCTA C	AAATAACAAGGCA TTGAAGAATGGCIT /AJGACGAGGGAA AGACGAAGGAAAG G	
1238	2875	3385	517	254	
cg43948629	cg43955093	cg43955093	cg43055918	cg43 <i>9</i> 74592	
159	160	161	162	163	

22 (22q13.1)	3 (3421)	7 (7p15)	
0	0		0
Human Gene SWISSPROT- ACC:P13866 SODIUM/GLUCOSE COTRANSPORTER 1 (NA(+)/GLUCOSE COTRANSPORTER 1) (HIGH AFFINITY SODIUM-GLUCOSE COTRANSPORTER 1) + Homo sapiens (Human), 664 aa.	Human Gene SWISSNEW- ACC:P00450 CERULOPLASMIN PRECURSOR (EC 1.16.3.1) (FERROXIDASE) - Homo sapiens (Human), 1065 aa.	Human Gene SWISSPROT- ACC:P41250 GLYCYL-TRNA SYNTHETASE (EC 6.1.1.14) (GLYCINE-TRNA LIGASE) (GLYRS) - Homo sapiens (Human), 685 aa.	Human Gene SWISSPROT- ACC:P02771 ALPHA- FETOPROTEIN PRECURSOR (ALPHA-FETOGLOBULIN) (ALPHA-1- FETOPROTEIN) - Homo sapiens (Human), 609 aa.
UNCLASSIFIE D	UNCLASSIFIE D	UNCLASSIFIE D	UNCLASSIFIE D
SILENT- CODING	SILENT: CODING	SILENT.	SILENT- CODING
Asp	Lys	Ala	Ojn Ojn
Asp	Lys	Ala	Olla
[O	∢	Ð
		E-4	⋖
AAGGACGCAACGC TGCCACCATGGA[C /T]AGTAGCACCTG GAGCCCCAAGACC A	TGAAAGTATTCAA TCCCAGAAGGAAI A/G]CTGGAATTTG CCCTTCTGTTTCTA G	GCTGGCGCACTGC TAGCCTCAGAGG[T /A]GCCAGCACCTC CTCAGCCCCCGCG C	AAAACCAGCTACC TGCCTTTCTGGA{A/ GJGAACTTTGCCAT GAGAAAGAAATTT
206	2757	2472	481
cg43956384	cg44025634		cg44024279
164	165	166	167

41	X (Xq28)	I (1q42)		2 (2cen)
5.00E-289	1.80E-287 X	3.90E-257	6.90E-239	3.00E-227 2
Human Gene SWISSNEW- ACC:Q13573 NUCLEAR PROTEIN SKIP (SNW1 PROTEIN) (NUCLEAR RECEPTOR COACTIVATOR NCOA-62) - Homo sapiens (Human), 536 aa.	Human Gene SWISSPROT- ACC:P51854 TRANSKETOLASE 2 (EC 2.2.1.1) (TK 2) (TRANSKETOLASE RELATED PROTEIN) - Homo sapiens (Human), 557 aa.	Human Gene SWISSPROT- ACC:P01019 ANGIOTENSINOGEN PRECURSOR - Homo sapiens (Human), 485 aa.	Human Gene SPTREMBL- ACC:043411 HYPOTHETICAL 49.3 KD PROTEIN - HOMO SAPIENS (HUMAN), 442 aa (fragment).	Human Gene SWISSPROT- ACC:P09529 INHIBIN BETA B CHAIN PRECURSOR (ACTIVIN BETA-B CHAIN) - Homo sapiens (Human), 407 aa.
UNCLASSIFIE D	UNCLASSIFIE D	UNCLASSIFIE D	UNCLASSIFIE D	UNCLASSIFIE D
SILENT- CODING	SILENT. CODING	SILENT- CODING	SILENT- CODING	SILENT- CODING
Glu	Gin	Gly	Arg	, lb
ng O	n _l g	Gly	Arg	Gly
<u></u>	<u>o</u>	<u> </u>	E	O
ပ	K	O	¥	U
CATGAGTTTTGATC CCAGCTCTTCT[CT] JTCCCTGGCTTTCT GGGCCATTTCTC	TTGGAGCTGGAAT TACTGTGTATGA[A/ GJGCCTTAGCAGCT GCTGATGAGCTTT	TCAACACCTACGT CCACTTCCAAGGG //JAAGATGAAGGG CTTCTCCCTGCTGG	TGGCTTGCACAAA TTGCTTGAAGAC(A AJCGATCCATGTAA GTGGACTGTCTTG	CTGGGCAGCTGCC CTCACAGTAGTT[C/ GJCCGTAGTAGCCG GTGGGTGCTATGA
1122	1778	1242	770	2186
cg43926814	cg40918088	cg43966985		cg42913861
168	169	170	171	172

2 (2cen)	12	12	(11p15.5
1	2.40E-225	2.40E-225	2.10E-224
Human Gene SWISSPROT- ACC:P09529 INHIBIN BETA B CHAIN PRECURSOR (ACTIVIN BETA-B CHAIN) - Homo sapiens (Human), 407 aa.	Human Gene SWISSPROT- ACC:P29080 (2- SYOLIGOADENYLATE SYNTHETASE 1B (EC 2.7.7) ((2-5')OLIGO(A) SYNTHETASE 1B) (2-5A SYNTHETASE 1B) - Mus musculus (Mouse), 414 aa.	Human Gene SWISSPROT- ACC:P29080 (2'- S')OLIGOADENYLATE SYNTHETASE 1B (EC 2.7.7) ((2-5')OLIGO(A) SYNTHETASE 1B) (2-5A SYNTHETASE 1B) - Mus musculus (Mouse), 414 aa.	Human Gene SWISSPROT- ACC:P04177 TYROSINE 3- MONOOXYGENASE (EC I.14.16.2) (TYROSINE 3- HYDROXYLASE) (TH) - Raftus norvegicus (Rat), 498 aa.
UNCLASSIFIE D	UNCLASSIFIE D	UNCLASSIFIE D	UNCLASSIFIE D
SILENT- CODING	SILENT- CODING	SILENT- CODING	SILENT: CODING
Arg	Gln	Gln	Thr
Arg	ulD	Gln	Th.
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o	U	U	ڻ ن
GCCGAGCCTGCAC CACCACAAAGGG CTJCGGTGCGACTC TTCGCCTGGGTCCA	CATAGAAGGCCAG GAGTCAGGAGACI CTJTGGGTTCTGTC CTGGATTATACAC C	GGAGTCAGGAGAC CTGGGTTCTGTC(C/ TJTGGATTATACAC CAGCTCACTGAGG	CCATGCCCACCCC CCACGCACGCACGCACGCACACGCCACAGGCCAAAGGCCAAGGCCAAGGCCAAGGCCAAGGCTTCCGCAGGGGGGGG
2354	256	268	53
c <u>g</u> 42913861	cg43929685	cg43929685	cg43918561
173	174	175	176

8 (8p12)	I	19 (19q13)	19 (19q13)
3.90E-218	4.90E-211	1.30E-210	1.30E-210
Human Gene SWISSPROT- ACC.P14902 INDOLEAMINE 2,3-DIOXYGENASE (EC 1.13.11.42) (IDO) (INDOLEAMINE- PYRROLE 2,3- DIOXYGENASE) - Homo sapiens (Human), 403 aa.	Human Gene SPTREMBL- ACC:Q99816 TUMOR SUSCEPTIBILITY PROTEIN - HOMO SAPIENS (HUMAN), 390 aa.	Human Gene SWISSPROT- ACC:015382 BRANCHED- CHAIN AMINO ACID AMINOTRANSFERASE, MITOCHONDRIAL PRECURSOR (EC 2.6.1.42) (BCAT(M)) - Homo sapiens (Human), 392 aa.	Human Gene SWISSPROT- ACC:015382 BRANCHED- CHAIN AMINO ACID AMINOTRANSFERASE, MITOCHONDRIAL PRECURSOR (EC 2.6.1.42) (BCAT(M)) - Homo sapiens (Human), 392 aa.
UNCLASSIFIE D	UNCLASSIFIE D	UNCLASSIFIE D	UNCLASSIFIE D
SILENT. CODING	SILENT:	SILENT. CODING	SILENT- CODING
Val	Ile	Leu	His
Val	Ile I	Leu	His
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¥	o O	U	ပ
ATTTAATGAATTTC CTGAAGACTGT[A/ GJAGAAGTACAACT GAGAAATCCCTTT	AAAACAATGATAT CGATGAAGTTAT[C /TJATTCCCACAGCT CCCTTATACAAAC	CACCATGAAGCAG TTGCTGCGGGCC[C/ TJTGGAGGAGGGCC GCGTGCGGGAAGT	TCCTGTACAAAGA CAGGAACCTCCA[C MJATTCCCACCATG GAAAATGGGCCTG
1885	1146	979	1074
-	cg43956382		cg43984681
177	8/1		180

- .	7.7	6
6.20E-204	1.90E-200	5.60E-191
SILENT- UNCLASSIFIE Human Gene SPTREMBL- CODING D ACC:P78545 ESE-1B - HOMO SAPIENS (HUMAN), 371 aa.	Human Gene SPTREMBL- ACC:060704 TYROSYLPROTEIN SULFOTRANSFERASE-2 - HOMO SAPIENS (HUMAN), 377 aa.	Human Gene SWISSNEW- ACC:Q03385 GUANINE NUCLEOTIDE DISSOCIATION STIMULATOR RALGDS FORM A (RALGEF) - Mus musculus (Mouse), 852 aa.
UNCLASSIFIE D	UNCLASSIFIE D	UNCLASSIFIE D
SILENT.	SILENT- CODING	SILENT- CODING
Pro	Leu	Leu
Pro	Leu	Leu
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1	ا ن	U
CTGCGGTGGAGAC A GTCAGAGCTGCCIA /GJGGGGAGGGGC TCCTGCGCCACAG C	ACCAGCTGCTCGT AGTACACAGGCA[G/A]GCACTTCTCCT TGCCTACCTCCATG	CTACCGCCAACTA TGACTTTGTCCT[C/ G]AAGAAGCGGAC CTTCACCAAGGGA G
	888	1114
cg43950996 1762	cg44024506	cg43980381
181	182	183

		9 (9qżz.2)
2.00E-189	1.40E-188	3.50E-178
DE 2- SE 2 L (2) UP		Human Gene SWISSPROT- ACC:P09467 FRUCTOSE-1,6- BISPHOSPHATASE (EC 3.1.3.11) (D-FRUCTOSE-1,6- BISPHOSPHATE 1- BISPHOSPHATE 1- PHOSPHOHYDROLASE) (FBPASE) - Homo sapiens (Human), 337 aa.
UNCLASSIFIE D	UNCLASSIFIE D	UNCLASSIFIE D
SILENT.	SILENT.	SILENT- CODING
Asn	Val	Ala
Asn	Val	Ala
E	<u> </u>	4
O	ပ	9
TCCCCTGGCAGAA CTACCACCTGAA[C TJGACTGGATGGA GGAGGAATACCGC C	ACATCCAGGTGGT GTTCGACGCCGT[C/ TJACCGACATCATC ATTGCCAACAACC	CAGTGACGGCAGG GTCAAAGTCCTT[G/ AJGCGTAGCCCTCG TTAAGGCTGTAGA
201	1497	
	cg43249389	cg43946951
184	185	186

16 (16p13.1 1)	(8p23.1)			
9 5 G	∞ 85			
1.20E-161	5.40E-157	1.20E-154	9.60E-148	9.60E-148 `
Human Gene SWISSPROT- ACC:Q14894 MU-CRYSTALLIN HOMOLOG (NADP- REGULATED THYROID- HORMONE BINDING PROTEIN) - Homo sapiens (Human), 314 aa.	Human Gene SWISSNEW- ACC:P11245 ARYLAMINE N- ACETYLTRANSFERASE, POLYMORPHIC (EC 2.3.1.5) (PNAT) (NAT-2) (ARYLAMINE ACETYLASE) - Homo sapiens (Human), 290 aa.	Human Gene SPTREMBL- ACC:Q15729 THYROTROPH EMBRYONIC FACTOR - HOMO SAPIENS (HUMAN), 303 aa.	Human Gene Homologous to SWISSNEW-ACC:Q12846 SYNTAXIN 4 - Homo sapiens (Human), 297 aa.	Human Gene Homologous to SWISSNEW-ACC:Q12846 SYNTAXIN 4 - Homo sapiens (Human), 297 aa.
UNCLASSIFIE D	UNCLASSIFIE D	UNCLASSIFIE D	UNCLASSIFIE D	UNCLASSIFIE D
SILENT- CODING	SILENT- CODING	SILENT- CODING	SILENT: CODING	SILENT:- CODING
id.	Тут	<u>T</u>	Arg	Gly
Ħ	Tyr	崖	Arg	Gly
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5	O	U	ග	ප
AACCAGCCCACTG TGAGAAGACCAC[G/C]GTGTTCAAGTC TTTGGGAATGGCA G	CCACAATGTTAGG AGGGTATTTTA[C/ I]ATCCCTCCAGTT AACAAATACAGCA	CTGCCATCTTTCAG CCCTCTGAAAC CT JGTGTCCAGCACAG AATCTTCCCTGG	GCGCTTCCCAGGT CCGGACAATTCG[G /I]CAGACTATTGTC AAACTGGGGAATA	GGCAGCTGAAGAT CACCAATGCTGG[G /C]ATGGTGTCTGAT GAGGAGTTGGAGC
1054	482	499	350	701
cg43248117				cg43942977
187	88	189	190	191

	· · · · · · · · · · · · · · · · · · ·	∞	61
9.60E-148	5.10E-145	5.10E-145	2.80E-144
Human Gene Homologous to SWISSNEW-ACC:Q12846 SYNTAXIN 4 - Homo sapiens (Human), 297 aa.	Human Gene Homologous to SWISSNEW-ACC:P29218 MYO- INOSITOL-1(OR 4)- MONOPHOSPHATASE (EC 3.13.25) (IMP) (INOSITOL MONOPHOSPHATASE) (LITHIUM-SENSITIVE MYO- INOSITOL MONOPHOSPHATASE A1)- Homo sepiens (Human), 277 aa.	Human Gene Homologous to SWISSNEW-ACC:P29218 MYO- INOSITOL-1(OR 4)- MONOPHOSPHATASE (EC 3.1.3.25) (IMP) (INOSITOL MONOPHOSPHATASE) (LITHIUM-SENSITIVE MYO- INOSITOL MONOPHOSPHATASE A1) - Homo sapiens (Human), 277 aa.	Human Gene Homologous to SWISSPROT-ACC.P29692 ELONGATION FACTOR 1- DELTA (EF-1-DELTA) - Homo sapiens (Human), 281 aa.
UNCLASSIFIE D	UNCLASSIFIE D	UNCLASSIFIE D	UNCLASSIFIE D
SILENT- CODING	SILENT- CODING	SILENT. CODING	SILENT- CODING
Lea	Gly	Ser	Ala
Leu	Gly	Ser	Ala
H	O	E	<u>ප</u>
O	H	O	∢
GCGAGGTGTTTGT GTCCAATATCCT[G/ TJAAGGACACGCA GGTGACTCGACAG G	TGTACACTGCCAG AAAAGGAAAAGG T/GJGCCTTTTGTAA TGGTCAAAAACTA C	TCTTGGTGACTGA GTTGGGCTCTTC[C/ TJAGAACACCAGA GACTGTGAGAATG G	TAGAGCGCACACA A GGCCTCCAGCTG[A /G]GCCATGTCCGTC TCATCATCCCAAG
773	753	837	321
			cg43946394
192	193	194	195

8 (8q22)	2 (2p13)	20	19 (19q13.4)	61
]_	•		c ·	6.80E-95
Human Gene Homologous to SWISSPROT-ACC:P00915 CARBONIC ANHYDRASE I (EC 4.2.1.1) (CARBONATE DEHYDRATASE I) - Homo sapiens (Human), 260 aa.	Human Gene Homologous to SWISSPROT-ACC:Q05195 MAD PROTEIN (MAX DIMERIZER) - Homo sapiens (Human), 221 aa.	Human Gene Homologous to TREMBLNEW-ACC:AAD43195 PEROXISOMAL MEMBRANE PROTEIN PMP 24 - HOMO SAPIENS (HUMAN), 212 aa.	Human Gene Similar to TREMBLNEW-ACC:BAA13472 CD89_U08 - HOMO SAPIENS (HUMAN), 191 aa.	Human Gene Similar to TREMBLNEW-ACC:CAB43107 - PRENYLATED RAB ACCEPTOR 1 (PRA1) - HOMO SAPIENS (HUMAN), 185 aa.
UNCLASSIPIE D	UNCLASSIFIE D	UNCLASSIFIE D	UNCLASSIFIE D	UNCLASSIFIE D
SILENT- CODING	SILENT- CODING	SILENT- CODING	SILENT. CODING	SILENT- CODING
		ł		1
Phe	Ten	/aJ	Arg .	P. P.
Phe Phe	Leu Leu	Val	Arg Arg	Pro Pro
	Leu	k Va	Arg	
T.	Leu	k Va	G Arg	T Pro
CTGACAGCTACAG C T Phe GCTCTTTCAGTT[C/T]CATTTTCACTGG GGCAGTACAAATG	T G Leu	T C Val	A G	C T Pro
T Phe	AGAAGTTGAAGGG T G Leu GCTGGTGCCACT[T/ G]GGACCCGAATCA AGTCGACACACTA	TGGGTTCAGGGAT T C Val GTAGCCCTTCTC[T/ CJACAGCCAGGCG GCTCAGGGCAAAC A	GCCAATATAGGAT A G Arg AGGCACTACAG[A/GJTTCCGGTACA GTGACACCCTGGA GC	ACGGGGAGGACT C T Pro GCAGATGGAACC[C /TJGTGTGAGGTGTC TTCTGGGACCTGC

=		9 (12q23)	11	
5.10E-90	1.30E-89	3.80E-85	1.30E-77	5.60E-68
Human Gene Similar to SPIREMBL-ACC:014803 BCL- X/BCL-2 BINDING PROTEIN - HOMO SAPIENS (HUMAN), 168 aa (fragment).	Hunan Gene Similar to TREMBLNEW-ACC:BAA34941 HUMAN CMAP - HOMO SAPIENS (HUMAN), 167 aa.	Human Gene Similar to SWISSPROT-ACC:P09496 CLATHRIN LIGHT CHAIN A (BRAIN AND LYMPHOCYTE LCA) - Homo sapiens (Human), 248 aa.	Human Gene Similar to SPTREMBL-ACC:000496 IPL (IPL) - HOMO SAPIENS (HUMAN), 152 aa.	Human Gene Similar to REMTREMBL-ACC:G36907 T- CELL RECEPTOR ALPHA- CHAIN HAPS8 V(A)10.1-1(A)T - HOMO SAPIENS (HUMAN), 135 aa (fragment).
UNCLASSIFIE D	UNCLASSIFIE D	UNCLASSIFIE D	UNCLASSIFIE D	UNCLASSIFIE D
SILENT- CODING	SILENT. CODING	SILENT. CODING	SILENT- CODING	SILENT- CODING
Arg	Leu	Pro	Leu	Arg
Arg		Pro	Leu	Arg
4	U	ပ	⊢	_ව
O	<u>ල</u>	1	ပ	V
AGAGGTTGGGGGG CGCCGAGCGCGAI G/A]CGGCCCCGAA AGGGGCTGGGCTC CT	TAGTGAAAGGCCT GAAATATATGCT[G /C]GAGGTGGAAAT TGGCAGAACTACC T	CTCCATCAACAGC ATCCGGACTGCA T /CJGGCGGCTCGCC OTGCGGCTGGGGC C	CGACGAGGTGCTA CGCGAGGGCGAG[CTJTGGAGAAGCG CAGCGACAGCCTC TJ	TTTTTCCAGCTTA CAATGGTACAG[A/ GCAGGAGCCTGG GGAAGGTCCTGTC C
871	682		136	162
				cg1527767
201	202			205

11 (11p15.2)	7 (7q3S)			17
5.10E-58		1.60E-54	2.00E-54	0
Human Gene Similar to SWISSNEW-ACC:P06881 CALCITONIN GENE-RELATED PEPTIDE I PRECURSOR (CGRP- I) (ALPHA-TYPE CGRP) - Homo sapiens (Human), 128 aa.	Human Gene Similar to REMTREMBL-ACC:D1002898 T- CELL RECEPTOR BETA-CHAIN V REGION - HOMO SAPIENS (HUMAN), 112 aa (fragment).	Human Gene Similar to REMTREMBL-ACC:G33S09 T CELL RECEPTOR - HOMO SAPIENS (HUMAN), 118 aa (fragment).	Human Gene Similar to SWISSNEW-ACC:P47992 LYMPHOTACTIN PRECURSOR (CYTOKINE SCM-1) (ATAC) (LYMPHOTAXIN) (SCM-1- ALPHA) - Homo sapiens (Human), 114 aa.	ATPase_associat Human Gene SPTREMBL- ed D:Q93050 VACUOLAR-TYPE H(+)-ATPASE 115 KDA SUBUNIT - HOMO SAPIENS (HUMAN), 831 aa.
UNCLASSIFIE D	UNCLASSIFIE D	UNCLASSIFIE D	UNCLASSIFIE D	ATPase_associat ed
SILENT- CODING	SILENT: CODING	SILENT: CODING	SILENT- CODING	SILENT: NONCOD ING
Thr	Ten	Ħ.	HE	
Thr	ne	FL.	Thr	
Т	U	ပ	<u>o</u>	gap
U	⊢	H	4	_ව
AGAAGAGACCTG TGACACTGCCAC[C MJTGTGTGACTCAT CGGCTGGCAGGCT	TCATCCTGAGITICT AAGAAGCTCCT[I/ C]CTCAGTGACTCT GGCTTCTATCTCT	CTCTGGTTGTCCAC GAGGGAGACACTY CJGTAACTCTCAAT TGCAGTTATGAAG	AAGTCTGTGCTGA TCCACAAGCCAC[A /G]TGGGTGAGAGA CGTGGTCAGGAGC A	AGGGAGGCGGGGA GGGTAGCATGGGI G/gap]CACACGGCC CTCACAGGGACTC ACT
316	300	317	249	1571
cg40968986	cg42550133	og2526759	cg41664708	cg43300673
206	207	208	209	210

				
9	=	m	E	m
4.00E-121	9.40E-58	0	0	0
ATPase associat Human Gene Homologous to SPTREMBL-ID:Q18788 C52E4.5 - CAENORHABDITIS ELEGANS, 590 aa.	ATPase_associat Human Gene Similar to SPTREMBL-ID:Q15332 GAMMA SUBUNIT OF SODIUM POTASSIUM ATPASE LIKE - HOMO SAPIENS (HUMAN), 126 aa.	Human Gene SWISSPROT- ID:P18084 INTEGRIN BETA-5 SUBUNIT PRECURSOR - HOMO SAPIENS (HUMAN), 799 aa.	Human Gene SWISSPROT- ID:P18084 INTEGRIN BETA-5 SUBUNIT PRECURSOR - HOMO SAPIENS (HUMAN), 799 aa.	Human Gene SWISSPROT- ID:P18084 INTEGRIN BETA-5 SUBUNIT PRECURSOR - HOMO SAPIENS (HUMAN), 799 aa.
	ATPase_associat	cadherin	cadherin	cadherin
SILENT- NONCOD ING	SILENT- NONCOD ING	SILENT- NONCOD ING	SILENT- NONCOD ING	SILENT: NONCOD ING
4	U	[F	ප
gap	¥	Ø	ග	gap
AGTTGAAATCAGA GAGGAATAAAAIB ap/AJGACATTTTAT ATTTTATTTTTTTTCTGCTC C	TAAGCATGAGGTG GCACGAGGCAGG[A/C]GTTGGCGATG CCACCTGGGGGTC AC	GGTCCCCTTGCTTT ATCCCAAGCTC[G/T JGAGGGACGCAGC CTGGCATGGCTCT	GTCCCCTTGCTTTA TCCCAAGCTCG[G/T JAGGGACGCAGCCT GGCATGGCTCTG	CTTTATCCCAAGCT CGGAGGGACGC[ga p/G]AGCCTGGCATG GCTCTGGCCTAGC A
2570	961	909	209	615
cg43284434	cg43132502	cg43931765	cg43931765	cg43931765
211	212	213	214	215

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2	3	რ	<u>м</u>	m .
0	0	0	0	0
Human Gene SWISSPROT- ID:P18084 INTEGRIN BETA-5 SUBUNIT PRECURSOR - HOMO SAPIENS (HUMAN), 799 aa.	Human Gene SWISSPROT- ID:P18084 INTEGRIN BETA-5 SUBUNIT PRECURSOR - HOMO SAPIENS (HUMAN), 799 aa.	Human Gene SWISSPROT- ID:P18084 INTEGRIN BETA-5 SUBUNIT PRECURSOR - HOMO SAPIENS (HUMAN), 799 aa.	Human Gene SWISSPROT- ID:P18084 INTEGRIN BETA-5 SUBUNIT PRECURSOR - HOMO SAPIENS (HUMAN), 799 aa.	Human Gene SWISSPROT- ID:P18084 INTEGRIN BETA-5 SUBUNIT PRECURSOR - HOMO SAPIENS (HUMAN), 799 aa.
cadherin	cadherin	cadherin	cadherin	cadherin
SILENT- NONCOD ING	SILENT- NONCOD ING	SILENT- NONCOD ING	SILENT- NONCOD ING	SILENT: NONCOD ING
H	E →	H	F	<u>[</u>
gap	gap	gap.	gap	gap
TAGCAGCCAGGTG ACATGGCCAGGC[g ap/T]ACCTTCCTGT ACAGGCACTGTGG GC	GCCAGGTGACATG GCCAGGCACCTT[g ap/T]CCTGTACAGG CACTGTGGGCTCCT G	AGGTGACATGGCC g AGGCACCTTCCT[ga p/T]GTACAGGCACT GTGGGCTCCTGGC C	AGGTGACATGGCC g AGGCACCTTCCT[ga p/TjGTACAGGCACT GTGGGCTCCTGGC C	AGGTGACATGGCC A AGGCACCTTCCT[ga p/T]GTACAGGCACT GTGGGCTCCTGGC C
099	965	899	899	899
cg43931765	cg43931765	cg43931765	cg43931765	cg43931765
216	217	218	219	

	16 (16p11.2)	1 (1923)
19	(16	D 1
		I.00E-218
0	0	00.1
Human Gene SPTREMBL- ID:Q15065 OB-CADHERIN-1 - HOMO SAPIENS (HUMAN), 796 aa.	Human Gene SWISSPROT- ID:P20701 LEUKOCYTE ADHESION GLYCOPROTEIN LFA-1 ALPHA CHAIN PRECURSOR (LEUKOCYTE FUNCTION ASSOCIATED MOLECULE 1, ALPHA CHAIN) (CD11A) (INTEGRIN ALPHA-L) - HOMO SAPIENS (HUMAN), 1170 aa.	Human Gene SWISSPROT- ID:P14151 L-SELECTIN PRECURSOR (LYMPH NODE HOMING RECEPTOR) (LEUKOCYTE ADHESION MOLECULE-1) (LAM-1) (LEUKOCYTE SURFACE ANTIGEN LEU-8) (TQ1) (GP90- MEL) (LEUKOCYTE- ENDOTHELIAL CELL ADHESION MOLECULE 1) (LECAMI) (CD62L) - HOMO SAPIENS (HUMAN), 372 aa.
cadherin	cadherin	cadherin
SILENT. NONCOD ING	SILENT- NONCOD ING	SILENT. NONCOD ING
O	T.	O
₹	5	Ŀ →
AATCCACAATCGG CATCAGGAAGCC[A /C]AAGTCCCAGTG GCCATTAGGGTCC T	TCCCTATGAGCCTG CAAAGGAGACA[G/ TJTCAGGAATGAGT TCCATGTTCGAGA	CAGTGCATCTGGG AAGATTCTACC[7/ CJGACCAACAGTTC CTTCAGCTTCCAT
4769	1406	1463
cg43952088	cg44010957	cg43956560
221	222	223

1 (1923)	1 (1923)
1.00E-218	1.00E-218
Human Gene SWISSPROT- ID:P14151 L-SELECTIN PRECURSOR (LYMPH NODE HOMING RECEPTOR) (LEUKOCYTE ADHESION MOLECULE-1) (LAM-1) (LEUKOCYTE SURFACE ANTIGEN LEU-8) (TQ1) (GP90- MEL) (LEUKOCYTE- ENDOTHELIAL CELL ADHESION MOLECULE 1) (LECAMI) (CD62L) - HOMO SAPIENS (HUMAN), 372 aa.	Human Gene SWISSPROT- ID:P14151 L-SELECTIN PRECURSOR (LYMPH NODE HOMING RECEPTOR) (LEUKOCYTE ADHESION MOLECULE-1) (LAM-1) (LEUKOCYTE SURPACE ANTIGEN LEU-8) (TQ1) (GP90- MEL) (LEUKOCYTE- ENDOTHELIAL CELL ADHESION MOLECULE 1) (LECAMI) (CD62L) - HOMO SAPIENS (HUMAN), 372 aa.
cadherin	cadherin
SILENT- NONCOD ING	SILENT: NONCOD ING
⋖	· · · · · · · · · · · · · · · · · · ·
O	U
CAACAGTTCCTTCA GCTTCCATTTC[G/A JCCCTCATTTATC CCTCAACCCCCA	TGCTCTCTTTCCC CTGCCCCAGA[C/ A]CTTTTATCCACT TACCTAGATTCTA
1492	2242
cg43956560	cg43956560
224	225

		T		
		·	5 (5p13)	9 (9q34.3)
4,10E,183		0	0	1.40E-104
Human Gene SWISSPROT- ID:P43235 CATHEPSIN K PRECURSOR (EC 3.4.22.38) (CATHESPIN O) (CATHEPSIN X) (CATHEPSIN O2) - HOMO SAPIENS (HUMAN), 329 aa.	Human Gene SWISSPROT- D:P27658 COLLAGEN ALPHA 1(VIII) CHAIN PRECURSOR (ENDOTHELIAL COLLAGEN) - HOMO SAPIENS (HUMAN), 744 aa.	Human Gene SWISSPROT- ID:P27658 COLLAGEN ALPHA 1(VIII) CHAIN PRECURSOR (ENDOTHELIAL COLLAGEN) - HOMO SAPIENS (HUMAN), 744 aa.	Human Gene SWISSPROT- ID:P10643 COMPLEMENT COMPONENT C7 PRECURSOR - HOMO SAPIENS (HUMAN), 843 aa.	Human Gene Homologous to SWISSPROT-ID:P07360 COMPLEMENT C8 GAMMA CHAIN PRECURSOR - HOMO SAPIENS (HUMAN), 202 aa.
cathepsin	collagen	collagen	complement	complement
SILENT- NONCOD ING	SILENT- NONCOD ING	SILENT- NONCOD ING	SILENT- NONCOD ING	SILENT: NONCOD ING
<	ڻ ٽ	Ö	U U	ڻ ن
 	U	U		
TGGCCACAGTGAA AAAGGTCATGGG[T A]GGAGAGAAGCA AAGTAGGAAGGAT C	ACCGCACCCTTTCC ACCGGTGGGGG[C/ G]CCCAGTGAAGTT TAACAAACTGCTG		GAAACCCAGTAGG // CTCCTGGAGGCC[A /C]TGGTCAGCTTGC TTGGAATCCAGCA	TGGTGGTGCTACC C CTTGGCCTCCCA[C/ G]AGTCCTGCCACC CTGCTGCCACC
428	1972	2096	2546	64
cg43264626	cg43011543	cg43011543	cg439337 <i>57</i>	cg41553795
226	227	228	229	230

3 (3q26.3)	3 (3q26.3)	17 (17q11.2)
1.20E-189	1.20E-189	1.50E-107
Human Gene SWISSPROT- ID-40225 THROMBOPOIETIN PRECURSOR (MEGAKARYOCYTE COLONY STIMULATING FACTOR) (C- MPL LIGAND) (ML) (MEGAKARYOCYTE GROWTH AND DEVELOPMENT FACTOR) (MGDF) - HOMO SAPIENS (HUMAN), 353 aa.	Human Gene SWISSPROT- ID:P40225 THROMBOPOIETIN PRECURSOR (MEGAKARYOCYTE COLONY STIMULATING FACTOR) (C- MPL LIGAND) (ML) (MEGAKARYOCYTE GROWTH AND DEVELOPMENT FACTOR) (MGDF) - HQMO SAPIENS (HUMAN), 3\$3 aa.	Human Gene Homologous to SWISSPROT-ID:P09919 GRANULOCYTE COLONY- STIMULATING FACTOR PRECURSOR (G-CSF) (PLURIPOIETIN) - HOMO SAPIENS (HUMAN), 207 aa.
Jso	csf	ુ કર ે
SILENT- NONCOD ING	SILENT- NONCOD ING	SILENT- NONCOD ING
	<u> </u>	
E	T	F
o	U	U
AGCCCTTCTCCACC CGGATAGATTC[C/T JTCACCCTTGGCCC GCCTTTGCCCCA	ACCCGGATAGATT CCTCACCCTTGG[C/ TJCCGCCTTTGCCC CACCCTACTCTGC	GTGCCTGGACATTT C GCCTTGCTGGA[CT JGGGGACTGGGGA TGTGGGAGGGAGC
168	179	1356
cg42542496	cg42542496	cg41533258
231	232	233

	
რ	01
1.10E-77	5.80E-303
Human Gene Similar to SWISSNEW-ID:P08700 INTERLEUKIN-3 PRECURSOR (IL-3) (MULTIPOTENTIAL COLONY-STIMULATING FACTOR) (HEMATOPOIETIC GROWTH FACTOR) (P-CELL STIMULATING FACTOR) (MAST-CELL, GROWTH FACTOR) (MCGF) - HOMO SAPIENS (HÜMAN), 152 aa. pcls:SWISSPROT-ID:P08700 INTERLEUKIN-3 PRECURSOR (IL-3) (MULTIPOTENTIAL COLONY-STIMULATING FACTOR) (HEMATOPOIETIC GROWTH FACTOR) (MAST-CELL GROWTH FACTOR) (MCGF) - HOMO SAPIENS (HÜMAN), 152 aa.	Human Gene SWISSPROT- ID:P00367 GLUTAMATE DEHYDROGENASE 1 PRECURSOR (EC 1.4.1.3) (GDH) - HOMO SAPIENS (HUMAN), 558 aa.
jso	dehydrogenase
SILENT- NONCOD ING	SILENT- NONCOD ING
	·
O .	O
gap p	5
ACGACTTTGAGCC TCGCGATCTTT[ga p/G]AGTCCAACGTC CAGCTCGTTCTTG	TGCGGCTTAAAAG GGCAACCCGCGG (CJGGACCCTTCCTC CCTAGTCGCGGGG
657	225
	cg44036323
234	235

7 (7431)	4 (4q22)	
5.10E-272	1.30E-209	·
Human Gene SPTREMBL- ID:Q14131 DIHYDROL.POAMIDE DEHYDROGENASE - HOMO SAPIENS (HUMAN), 511 aa.	Human Gene SWISSNEW- ID:P08319 ALCOHOL DEHYDROGENASE CLASS II PI CHAIN (EC 1.1.1) - HOMO SAPIENS (HUMAN), 391 aa.lpcls:SWISSPROT-ID:P08319 ALCOHOL DEHYDROGENASE CLASS II PI CHAIN (EC 1.1.1) HOMO SAPIENS (HUMAN), 391 aa.	Human Gene SWISSNEW- ID:P19838 NUCLEAR FACTOR NF-KAPPA-B P105 SUBUNIT (DNA-BINDING FACTOR KBF1) (EBP-1) [CONTAINS: NUCLEAR FACTOR NF-KAPPA-B P50 SUBUNIT-HOMO SAPIENS (HUMAN), 969 aa. pcls:SWISSPROT-ID:P19838 NUCLEAR FACTOR NF-KAPPA-B P105 SUBUNIT (CONTAINS: NUCLEAR FACTOR NF-KAPPA-B P105 SUBUNIT (CONTAINS: NUCLEAR FACTOR NF-KAPPA-B P105 SUBUNIT) (DNA-BINDING FACTOR KBF1) (EBP-1)-HOMO SAPIENS (HUMAN), 969 aa.
dehydrogenase	dehydrogenase	dna_ma_bind
SILENT. NONCOD ING	SILENT- NONCOD ING	SILENT- NONCOD ING
H	U	ပ
υ	dag	d සිති
GAGAGACCATTTA CTTACATCAGTT[C/ TJGGTTTATAGACA TTTGAATCATATC	AGTTTCATTATACT TTTCTCTCCAC[gap/ G]TTTGTCTATGTT GAAAATTTTCTG	ACAAGACAGAGC TGAAGTGCATCC[g ap/C]AAAGGTGCTC AGAGAGCCGGCCC GC
766	995	3691
	cg43057018	cg44005808
236	237	

	Т	
	10	10
0	1.40E-159	1.40E-159
Human Gene SWISSNEW- ID:P19838 NUCLEAR FACTOR NF-KAPPA-B P105 SUBUNIT (DNA-BINDING FACTOR KBF1) (EBP- 1) [CONTAINS: NUCLEAR FACTOR NF-KAPPA-B P50 SUBUNIT] - HOMO SAPIENS (HUMAN), 969 aa.jpcis:SWISSPROT-ID:P19838 NUCLEAR FACTOR NF-KAPPA-B P105 SUBUNIT (CONTAINS: NUCLEAR FACTOR NF-KAPPA-B P105 SUBUNIT (CONTAINS: NUCLEAR FACTOR NF-KAPPA-B P105 SUBUNIT) (DNA-BINDING FACTOR KBF1) (EBP-1) - HOMO SAPIENS (HUMAN), 969 aa.	Human Gene SPTREMBL- ID: Q99612 DNA-BINDING PROTEIN CPBP - HOMO SAPIENS (HUMAN), 290 aa (fragment).	Human Gene SPTREMBL- ID:Q99612 DNA-BINDING PROTEIN CPBP - HOMO SAPIENS (HUMAN), 290 aa (fragment).
dna_ma_bind	dna ma bind	dna ma bind
SILENT. NONCOD ING	SILENT- NONCOD ING	SILENT: NONCOD ING
o .	Ą	[-
de 89	dea	фар
TCTTCCTTCTCCAG CCGGCAGGCCC[gap /G]CGCCGCTTAGG AGGGAGAGCCCAC C	TGGCGAGTCCAGG GTCACCCACATA[g ap/a]CCATGCACCA CGGGTGCTATGCC GC	GAGTCCAGGGTCA CCCACATACCAT[ga p/T]GCACCACGGGT GCTATGCCGCTTCT
930	1244	1248
cg44005808	cg43956159	cg43956159
239	240	241

			1936.13
2	01	01	(3p)
1.40E-159	1.40E-159	1.40E-159	1.30E-60
Human Gene SPTREMBL- ID:Q99612 DNA-BINDING PROTEIN CPBP - HOMO SAPIENS (HUMAN), 290 aa (fragment).	Human Gene SPTREMBL- ID:Q99612 DNA-BINDING PROTEIN CPBP - HOMO SAPIENS (HUMAN), 290 aa (fragment).	Human Gene SPTREMBL- ID:Q99612 DNA-BINDING PROTEIN CPBP - HOMO SAPIENS (HUMAN), 290 aa (fragment).	Human Gene Similar to SWISSNEW-ID:Q02535 DNA- BINDING PROTEIN INHIBITOR ID-3 (ID-LIKE PROTEIN INHIBITOR HLA 1821) (HELIX- LOOP-HELIX PROTEIN HEIR-1) - HOMO SAPIENS (HUMAN), 119 aa. [pcls:SWISSPROT- ID:Q02535 DNA-BINDING PROTEIN INHIBITOR ID-3 (ID- LIKE PROTEIN INHIBITOR HLH 1R21) (HELIX-LOOP- HELIX PROTEIN HEIR-1) - HOMO SAPIENS (HUMAN), 119 aa.
dna_ma_bind	dna_ma_bind	dna_ma_bind	dna_ma_bind_in hib
SILENT- NONCOD ING	SILENT. NONCOD ING	SILENT- NONCOD ING	SILENT- NONCOD ING
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TACCATGCACCAC GGGTGCTATGCC[G /A]CTTCTTACAGGA CCTTTTTAGCCCT	CCTGGAGGCAACT GGGTAGGGTGCA[G/CJAACGGCATGC TTTGGCTGGAACA CG	CAGAACGGCATGC TTTGGCTGGAAC[ga p/C]ACGCATCCCTC CTTCCACGGCCGG .C	CAGAGCTAGCTCT GGCTCTTCAGGC[C/ TJACAAGTTCACAG TCCTTCGCTCCTG
1268	1342	1364	
	cg43956159	cg43956159	cg43971258
242	243	244	245

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9 (9p24)	9 (9p24)	9 (9p24)
0	0	0
Human Gene SWISSPROT- ID: P98155 VERY LOW- DENSITY LIPOPROTEIN RECEPTOR PRECURSOR (VLDL RECEPTOR) - HOMO SAPIENS (HUMAN), 873 aa.	Human Gene SWISSPROT- ID:P98155 VERY LOW- DENSITY LIPOPROTEIN RECEPTOR PRECURSOR (VLDL RECEPTOR) - HOMO SAPIENS (H\$\\$MAN), 873 aa.	Human Gene SWISSPROT- ID:P98155 VERY LOW- DENSITY LIPOPROTEIN RECEPTOR PRECURSOR (VLDL RECEPTOR) - HOMO SAPIENS (HUMAN), 873 aa.
cbh	ebh	ьф
SILENT: NONCOD ING	SILENT: NONCOD ING	SILENT- NONCOD ING
U	<u>o</u>	U
dea	gap	•
C) AC)		CCTCCTTCTCCCCC TTTCCCCTCCCIAC JGCCCCCACCTTCT TCCTCCTTTCG
3373	3739	514
cg43982507		cg43982507
247	248	249
	cg43982507 3373 GATACCTTTGCGTG gap C SILENT- eph Human Gene SWISSPROT- 0 NONCOD RECEPTOR ID:P98155 VERY LOW- ID:P98155 VERY LOW-	cg43982507 3373 GATACCTTTGCGTG gap C SILENT- eph Human Gene SWISSPROT- 0 CGTCAAGCTTG[gap CGTGTACTTGACCG NONCOD DENSITY LIPOPROTEIN RECEPTOR PRECURSOR (VLDL RECEPTOR) - HOMO Cg43982507 3739 CAAAAAAAATTTAT gap G SILENT- eph Human Gene SWISSPROT- 0 P/GJTACGTATGAAT RONCOD DENSITY LIPOPROTEIN NONCOD DENSITY LIPOPROTEIN 0 RECEPTOR, PLOW- NONCOD DENSITY LIPOPROTEIN NONCOD DENSITY LIPOPROTEIN 0 RAACTAAAAAATTTTG[ga NONCOD DENSITY LIPOPROTEIN NONCOD DENSITY LIPOPROTEIN RAACTAAAAAAATTTTG[ga RECEPTOR, PRECURSOR (VLDL RECEPTOR, PROCURSOR (VLDL RECEPTOR, PROCURSOR RAPIENS (HUMAN), 873 aa. SAPIENS (HUMAN), 873 aa. SAPIENS (HUMAN), 873 aa.

11 (11923)	11 (11q23)	m
1.80E-203	1.80E-203	O
Human Gene SWISSNEW- ID:P06727 APOLIPOPROTEIN A- IV PRECURSOR (APO-AIV) - HOMO SAPIENS (HUMAN), 396 aa. pcls:SWISSPROT-ID:P06727 APOLIPOPROTEIN A-IV PRECURSOR (APO-AIV) - HOMO SAPIENS (HUMAN), 396 aa.	Human Gene SWISSNEW- ID:P06727 APOLIPOPROTEIN A- IV PRECURSOR (APO-AIV) - HOMO SAPIENS (HUMAN), 396 aa.pcls:SWISSPROT-ID:P06727 APOLIPOPROTEIN A-IV PRECURSOR (APO-AIV) - HOMO SAPIENS (HUMAN), 396 aa.	Human Gene SWISSPROT- ID:P51178 1- PHOSPHATIDYLINOSITOL-4,5- BISPHOSPHATE PHOSPHODIESTERASE DELTA 1 (EC 3.1.4.11) (PLC-DELTA-1) (PHOSPHOLIPASE C-DELTA-1) (PLC-III) - HOMO SAPIENS (HUMAN), 756 aa.
ųdo	ebh	esterase
SILENT- NONCOD ING	SILENT. NONCOD ING	SILENT. NONCOD ING
		,
Į	T	U
dag	da da	U
CTGCCCTGCCACCT GTCTGTCTGTG[gap/ TJCCAAAGAAGTTC TGGTATGAACTTG	CTGCCCTGCCACCT GTCTGTCTGTC[gap/ TJCCAAAGAAGTTC TGGTATGAACTTG	TCCCTCCAGGACT AGGCTGGAGGAAI G/CJCCAGTGGGGT CCCCCTGAGTGG GC
1371	1371	2376
cg41554010	cg41554010	cg43984905
250	251	252

		(11pter)
m .	<i>r</i>	
		1.80E-195
0 -5 -5 -C	0	
Human Gene SWISSPROT- ID:P51178 1- PHOSPHATIDYLINOSITOL-4,5- BISPHOSPHATE PHOSPHODIESTERASE DELTA 1 (EC 3.1.4.11) (PLC-DELTA-1) (PHOSPHOLIPASE C-DELTA-1) (PLC-III) - HOMO SAPIENS (HUMAN), 756 aa.	Human Gene SWISSPROT- ID:P08183 MULTIDRUG RESISTANCE PROTEIN 1 (P- GLYCOPROTEIN 1) - HOMO SAPIENS (HUMAN), 1280 aa.	Human Gene SWISSPROT- ID:P16070 CD44 ANTIGEN PRECURSOR (PHAGOCYTIC GLYCOPROTEIN I) (PGP-1) (HUTCH-I) (EXTRACELLULAR MATRIX RECEPTOR-III) (ECMR-III) (GP90 LYMPHOCYTE HOMING/ADHESION RECEPTOR) (HERMES ANTIGEN) (HYALURONATE RECEPTOR) (HERMES ANTIGEN) (HYALURONATE RECEPTOR) (HEPGRAN SULFATE PROTEOGLYCAN) (EPICAN) (CDW44) - HOMO SAPIENS (HUMAN), 742 aa.
esterase	glycoprotein	glycoprotein
SILENT- NONCOD ING	SILENT- NONCOD ING	SILENT- NONCOD ING
U	A	V
Ö	5	O
000		TTTCTAGAGGGGG C TCTGTTGAAGAT[G/ AJTGTAACTAGTAC ACCCCAACCCCCA
2440	382	
100	cg43992911	cg43932434
253	254	255

(11pter)	1 (1921)
1.80E-195	3.10E-185
Human Gene SWISSPROT- ID:P16070 CD44 ANTIGEN PRECURSOR (PHAGOCYTIC GLYCOPROTEIN I) (PGP-1) (HUTCH-I) (EXTRACELLULAR MATRIX RECEPTOR-III) (ECMR-III) (GP90 LYMPHOCYTE HOMING/ADHESION RECEPTOR) (HERMES ANTIGEN) (HYALURONATE RECEPTOR) (HERMES ANTIGEN) (HYALURONATE RECEPTOR) (HERMES ANTIGEN) (HYALURONATE RECEPTOR) (HERMES SAPIENS (HUMAN), 742 aa.	Human Gene SWISSNEW- ID:P15813 T-CELL SURFACE GLYCOPROTEIN CDID PRECURSOR (CDID ANTIGEN) (R3G1) - HOMO SAPIENS (HUMAN), 335 aalpols:SWISSPROT-ID:P15813 T-CELL SURFACE GLYCOPROTEIN CDID PRECURSOR (CDID ANTIGEN) (R3G1) - HOMO SAPIENS (HUMAN), 335 aa.
glycoprotein	glycoprotein
SILENT- NONCOD ING	SILENT. NONCOD ING
	;
o	_ნ
₹	Ą
CCCCAACCCCCAA CCTCAGTGGAAA[A /G]CAATGCCCAGG GATTAGGCTATGG A	GCGCAGGTCAGAG GGCGGCCGCAGCI A/GJGGCCTCCGCG AGGTCCCCACGCC GG
306	366
cg43932434	cg43318219
256	257

		6 (6p25)	6 (6p25)
8.20E-67	8.20E-67	8.90E-61	8.908-61
Human Gene Similar to SWISSPROT-ID:Q08878 FIBULIN-1, ISOFORM C PRECURSOR (BASEMENT- MEMBRANE PROTEIN 90) (BM- 90) - MUS MUSCULUS (MOUSE), 685 aa.	Human Gene Similar to SWISSPROT-ID:Q08878 FIBULIN-1, ISOFORM C PRECURSOR (BASEMENT- MEMBRANE PROTEIN 90) (BM- 90) - MUS MUSCULUS (MOUSE), 685 aa.	Human Gene Similar to SWISSPROT-ID:P49222 ERYTHROCYTE MEMBRANE PROTEIN BAND 4.2 (P4.2) (PALLIDIN) - MUS MUSCULUS (MOUSE), 690 aa.	Human Gene Similar to SWISSPROT-ID:P49222 ERYTHROCYTE MEMBRANE PROTEIN BAND 4.2 (P.4.2) (PALLIDIN) - MUS MUSCULUS (MOUSE), 690 aa.
glycoprotein	glycoprotein	glycoprotein	glycoprotein
SILENT- NONCOD ING	SILENT- NONCOD ING	SILENT- NONCOD ING	SILENT- NONCOD ING
,			
Вар	gap	П	П
[L	် ပ	A
CTCTATACTGTACA CTCACCCATAA[T/g ap]TCAAACAATTA CACCATGGTATAA A	TCTATACTGTACAC TCACCCATAATIT/g apjCAAACAATTAC ACCATGGTATAAA G	GCCGAATAGCCTG GGTTTGGAAAAG[C //JATGTTTTGAAA TATGTGGGATCTC	TACTGACCTAAAT CACACCCTAGACIA /TJTATCAGAGGA AATTCTGACCATA A
1954	1955		385
cg43967861	cg43967861	cg43965366	cg43965366
258	259	260	261

				
12	7	7	7	
3.30E-54	1.20 E-2 24	1.20E-224	1.20E-224	1.60E-206
Human Gene Similar to SWISSPROT-ID:P13983 EXTENSIN PRECURSOR (CELL WALL HYDROXYPROLINE- RICH GLYCOPROTEIN) - NICOTIANA TABACUM (COMMON TOBACCO), 620 aa.	Human Gene SWISSPROT- ID:P50219 HOMEOBOX PROTEIN HB9 - HOMO SAPIENS (HUMAN), 401 aa.	Human Gene SWISSPROT- ID:P50219 HOMEOBOX PROTEIN HB9 - HOMO SAPIENS (HUMAN), 401 aa.	Human Gene SWISSPROT- ID:PS0219 HOMEOBOX PROTEIN HB9 - HOMO SAPIENS (HUMAN), 401 aa.	Human Gene TREMBLNEW- ID:G2896172 LIM HOMEOBOX PROTEIN COFACTOR - HOMO SAPIENS (HUMAN), 373 aa.
glycoprotein	homeobox	homeobox	<u> homeobox</u>	homeobox
SILENT- NONCOD ING	SILENT. NONCOD ING	SILENT- NONCOD ING	SILENT- NONCOD ING	SILENT- NONCOD ING
				·
to	ڻ ت	O	[-	O .
₹	gap	døå	U	⊢
TGTCCTTGAAGAA CATGCACTTGGC[A /G]CGGATGGCACA AGCAAAATGGTAG A	CCCGCGCCCAGT AGGAGCCCCGCG[g ap/G]CCCAGCAGGT GCGGCGCCACGG AG	CCAGCAGGTGCGG CGCGCACGGAGC[g ap/G]CGCCGGCCGG CGGCTTCTCCCGG AG	TGAAACTTGAAAC CGCCTCTGGAGC[C /TJGCCATTCTGCAG AGTATTTGGAAAA	TCCAAGAAAGGGT CATGGAAGCTTAĮT /CJTGGGAATAATC CTCTCAATTAGAA A
1255	1397	1423	1817	939
				c <u>g</u> 43980506
262	263	264	265	266

10	16 (16q22)	16 (16q22)	7 (7p21)
1.30E-156	2.00E-220	2.00E-220	3.40E-108
Human Gene SWISSPROT- ID:P37980 INORGANIC PYROPHOSPHATASE (EC 3.6.1.1) (PYROPHOSPHATE PHOSPHO- HYDROLASE) (PPASE) - BOS TAURUS (BOVINE), 289 aa.	Human Gene SFTREMBL- D:Q13194 11-BETA- HYDROXYSTEROID DEHYDROGENASE TYPE 2 - HOMO SAPIENS (HUMAN), 405 aa.	Human Gene SPTREMBL- D:Q13194 11-BETA- HYDROXYSTEROID DEHYDROGENASE TYPE 2 - HOMO SAPIENS (HUMAN), 405 aa.	Human Gene Homologous to SWISSPROT-ID:P05231 INTERLEUKIN-6 PRECURSOR (IL-6) (B-CELL STIMULATORY FACTOR 2) (BSF-2) (INTERFERON BETA-2) (HYBRIDOMA GROWTH FACTOR) - HOMO SAPIENS (HUMAN), 212 aa.
hydrolase	hydroxy steroid	hydroxysteroid	interleukin
SILENT. NONCOD ING	SILENT: NONCOD ING	SILENT: NONCOD ING	SILENT- NONCOD ING
<u></u>	gap	deg	f _{ree}
_.	U	O	desa
6666667777777 TTTTTCCCG[6/f] TTTTTTTTTTTTTTTTTTTTTTTTTTTTTTTTTTTT	CTGGGGGTTTTCG GGGAGGAACCAA[G/gap]GGCTCACGG AGCCTCCTGTGCTG CA	GGGGGTTTTCGGG GAGGAACCAAGG[G/gap]CTCACGGAG CCTCCTGTGCTGCA GT	GAGTTAATTTATGT AAGTCATATTT[gap/ TJATATTTTAAGA AGTACCACTTGAA
100	503	505	1031
cg43961305	cg43998672	cg43998672	cg42908571
267	268	269	270

7 (7p21)	2 (2q35)	vo
3.40E-108	9.60E-191	1.60E-156
Human Gene Homologous to SWISSPROT-ID:P05231 INTERLEUKIN-6 PRECURSOR (IL-6) (B-CELL STIMULATORY FACTOR 2) (BSF-2) (INTERFERON BETA-2) (HYBRIDOMA GROWTH FACTOR) - HOMO SAPIENS (HUMAN), 212 aa.	interleukinrecept Human Gene (SWISSPROT- ID:P25025 HfGH AFFINITY INTERLEUKIN-8 RECEPTOR B (IL-8R B) (CXCR-2) (GRO/MGSA RECEPTOR) (IL-8 RECEPTOR TYPE 2) - HOMO SAPIENS (HUMAN), 360 aa.	Human Gene SWISSPROT- ID:P46926 PUTATIVE GLUCOSAMINE-6-PHOSPHATE ISOMERASE (EC 5.3.1.10) (GLUCOSAMINE-6- PHOSPHATE DEAMINASE) (OSCILLIN) (KIAA0060) - HOMO SAPIENS (HUMAN), 289 aa.
interleukin	interleukinrecept	isomerase
	SILENT- NONCOD ING	SILENT- NONCOD ING
[H	U	ڻ ت
da da da da da da da da da da da da da d	[-	4
CTTACCTCAAATA E AATGGCTAACTT[ga p/T]ATACATATTT TAAAGAAATATTT A	CAGCCCCATTGT GGTCACAGGAAGIT /CJAGAGGAGGCCA CGTTCTTACTAGTT	CCCAACCTGGGTTT GGCAGACATCA[A/ GJAATGATGGAGTA CATTTTGCAGATA
1178	1617	1133
cg42908571	cg42164914	cg43958501
271	272	273

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S	01	X (Xq21.3)
1.60E-156	0	0
Human Gene SWISSPROT- ID:P46926 PUTATIVE GLUCOSAMINE-6-PHOSPHATE ISOMERASE (EC 5.3.1.10) (GLUCOSAMINE- 6- PHOSPHATE DEAMINASE) (OSCILLIN) (KIAA0060) - HOMO SAPIENS (HUMAN), 289 aa.	Human Gene SWISSPROT- ID:Q04759 PROTEIN KINASE C, THETA TYPE (EC 2.7.1) (NPKC-THETA) - HOMO SAPIENS (HUMAN), 706 aa.	Human Gene SWISSPROT- D:Q06187 TYROSINE-PROTEIN KINASE BTK (EC 2.7.1.112) (BRUTON'S TYROSINE KINASE) (AGAMMAGLOBULINAEMIA TYROSINE KINASE) TYROSINE KINASE) (GELL PROGENITOR KINASE) (BPK) - HOMO SAPIENS (HUMAN), 659 aa
isomerase	kinase	kinase
SILENT. NONCOD ING	SELENT- NONCOD ING	SILENT- NONCOD ING
·		
4	o I	O
O	ļ.	· •
CACCCCCAGGTTCT C CCTAGTTCAGA[G/ A]AAAAGCTGTGA AAGTGGAAGAAGG A	TTTATTCTATTCCT ATCTGTGGATG[T/G]GTAAATGGCTGGG GGGCCAGCCCTG	AGCCTTTGTGCTCC CACTCAATACA[A/ CJAAAGGCCCCTCT CTACATCTGGGAA
802	2710	2259
cg43958501		cg42879455
274	275	276

X (Xq21.3)	6		6
0	1.405-290	1.40E-290	1.40E-290
Human Gene SWISSPROT- ID:Q06187 TYROSINE-PROTEIN KINASE BTK (EC 2.7.1.112) (BRUTON'S TYROSINE KINASE) (AGAMMAGLOBULINAEMIA TYROSINE KINASE) (TYROSINE KINASE) (CELL PROGENITOR KINASE) (BPK) - HOMO SAPIENS (HUMAN), 659 aa.	Human Gene SPTREMBL- ID: Q92749 TYPE 1 PHOSPHATIDYL,NOSITOL-4 PHOSPHATE 5-KINASE BETA (EC 2.7.1.68) (STM-7 PROTEIN)- HOMO SAPIENS (HUMAN), 540 aa.	Human Gene SPTREMBL- ID:Q92749 TYPE 1 PHOSPHATIDYLNOSITOL-4- PHOSPHATE 5-KINASE BETA (EC 2.7.1.68) (STM-7 PROTEIN) - HOMO SAPIENS (HUMAN), 540 aa.	Human Gene SPTREMBL- ID:Q92749 TYPE I PHOSPHATIDYLINOSITOL 4- PHOSPHATE 5-KINASE BETA (EC 2.7.1.68) (STM-7 PROTEIN) - HOMO SAPIENS (HUMAN), 540 aa.
kinase	kinase	kinase	kinase
SILENT- NONCOD ING	SILENT: NONCOD ING	SILENT- NONCOD ING	SILENT. NONCOD ING
O	E-	«	T
	O	O	des
AAAAAGGCCCCTC TCTACATCTGGG[A/ G]ATGCACCTCTTC TTTGATTCCCTGG	AGCAACTTGGCTG AGCCCACTACA[C //TACAGAGAAATC ATCAACCTGACTT A	TAAGAGTTTTCAA GATGTCAAACTT[C/ AJAGGCTGATCAGC AGATGGGATGTGA	TTTTTAAAAATCCA TCCACACACATIGap TIGGTAAATTAAG TATAAATTCTTTG
2283	2151	. 5200	2451
			cg43971741
277	278	279	280

	·			
61	119	16	71	
5.60E-267	5.60E-267	7.80E-173	1.40E-79	5.30E-55
Human Gene SWISSPROT. ID:P49840 GLYCOGEN SYNTHASE KINASE-3 ALPHA (EC 2.7.1.37) (GSK-3 ALPHA)- HOMO SAPIENS (HUMAN), 483 aa.	Human Gene SWISSPROT- ID:P49840 GLYCOGEN SYNTHASE KINASE-3 ALPHA (EC 2.7.1.37) (GSK-3 ALPHA) - HOMO SAPIENS (HUMAN), 483 aa.	Human Gene SPTREMBL- ID:Q15599 TYROSINE KINASE ACTIVATOR PROTEIN 1 (TKA- 1) - HOMO SAPIENS (HUMAN), 450 aa.	Human Gene Similar to SPTREMBL-ID:Q15599 TYROSINE KINASE ACTIVATOR PROTEIN I (TKA- 1) - HOMO SAPIENS (HUMAN), 450 aa.	Human Gene Similar to SWISSPROT-ID:P20505 30 KD PROTEIN KINASE HOMOLOG (EC 2.7.1) (PROTEIN B1) - VACCINIA VIRUS (STRAIN COPENHAGEN), 300 aa.
kinase	kinase	kinase	kinase	kinase
SILENT- NONCOD ING	SILENT- NONCOD ING	SILENT- NONCOD ING	SILENT- NONCOD ING	SILENT- NONCOD ING
Вар	ďeg	v	ပ	4
<u></u> ნ_	_ව	gap	¥	
AACGTCGATTCGC ACGTCCAACCT[G /gap]GCCCGCCCC TCCTACAGCTGTA AC	ACGTCGATTCGCA C CCGTCCAACCTG[G /gsp]CCCGCCCTC CTACAGCTGTAAC T	CACTTAATACCAG AGACCCCCCCE ap/CJTCCCCTCCCC CTTCCCCTCCCCT	AGACGTGTCTGCC ACAGGTCTCAGG[A /GJTAACAGATGCC CTGTCCACTGAGA G	TTTGATGGAAAGG TTGTCCACACTG[G/ A]GAATTATCACAC ACTTGATCAGGAA
1996	1997	1535	306	1876
cg43947749	cg43947749	cg44131752	cg43917718	cg43928048
281	282	283	284	285

9 (9p21)	19 (19q13.1)
2.60E-53	0
Human Gene Similar to SWISSPROT-ID:P42771 CYCLIN-DEPENDENT KINASE 4 INHIBITOR A (CDK41) (P16- INKA) (P16-INK4A) (MULTIPLE TUMOR SUPRESSOR 1) (MTS1) - HOMO SAPIENS (HUMAN), 156 aa.	Human Gene SWISSNEW- ID:P30530 TYROSINE-PROTEIN KINASE RECEPTOR UFO PRECURSOR (EC 2.7.1.112) (AXL ONCOGENE) - HOMO SAPIENS (HUMAN), 887 aa_lpcis:SWISSPROT-ID:P30530 TYROSINE-PROTEIN KINASE RECEPTOR UFO PRECURSOR (EC 2.7.1.112) (AXL ONCOGENE) - HOMO SAPIENS (HUMAN), 887 aa.
SILENT- Kinaseinhibitor NONCOD	kinasereceptor
SILENT- NONCOD ING	SILENT- NONCOD ING
×	0
U	A
CCCTCCGGATTCG GCGCGCTGCGG[C M]CCGCCGCGAGT GAGGGTTTTCGTG G	TCCAAGCTAAGCA / CTGCCACTGGGG[A /G]AAACTCCACCTT CCCACTTTCCCAC
	2943
cg42714751 208	cg43322545
286	287

19 (19q13.1)	19 (19q13.1)	16
0		0
Human Gene SWISSNEW- ID:P30530 TYROSINE-PROTEIN KINASE RECEPTOR UFO PRECURSOR (EC 2.7.1.112) (AXL ONCOGENE) - HOMO SAPIENS (HUMAN), 887 aa. [pcls::SWISSPROT-ID:P30530 TYROSINE-PROTEIN KINASE RECEPTOR UFO PRECURSOR (EC 2.7.1.112) (AXL ONCOGENE) - HOMO SAPIENS (HUMAN), 887 aa.	Human Gene SWISSNEW- ID:P30530 TYROSINE-PROTEIN KINASE RECEPTOR UFO PRECURSOR (EC 2.7.1.112) (AXL ONCOGENE) - HOMO SAPIENS (HUMAN), 887 aa.pcls:SWISSPROT-ID:P30530 TYROSINE-PROTEIN KINASE RECEPTOR UFO PRECURSOR (EC 2.7.1.112) (AXL ONCOGENE) - HOMO SAPIENS (HUMAN), 887 aa.	Human Gene SPTREMBL- ID:Q14807 KID (KINESIN-LIKE DNA BINDING PROTEIN) - HOMO SAP(ENS (HUMAN), 665 aa.
Kinasereceptor	kinasereceptor	kinesin
SILENT. NONCOD ING	SILENT- NONCOD ING	SILENT: NONCOD ING
<u>.</u>	O	5
	D	4 *
CCACCTCCATCCCA GACAGGTCCCT[C/ G]CCCTTCTCTGTG CAGTAGCATCACC		GTCTGATAGAAGA , GGAGCAGGAGAA[A/G]CAAATCGTTA AAACCTAGCGAAT TC
3037	3038	1040
cg43322545	og43322545	cg43980494
288	289	290

14	6 (6p21.3)	6 (6p21.3)	61	
1.90E-304	3.405-147	3.70E-134	1.80E-113	3.50E-81
Human Gene SWISSPROT- ID:Q07866 KINESIN LIGHT CHAIN (KLC) - HOMO SAPIENS (HUMAN), 569 aa.	Human Gene Homologous to SWISSPROT-ID:P13765 HLA CLASS II HISTOCOMPATIBILITY ANTIGEN, DO BETA CHAIN PRECURSOR - HOMO SAPIENS (HUMAN), 273 aa.	Human Gene Homologous to SWISSPROT-ID:P06340 HLA CLASS II HISTOCOMPATIBILITY ANTIGEN, DZ ALPHA CHAIN PRECURSOR (MHC DN-ALPHA) - HOMO SAPIENS (HUMAN), 250 aa.	Human Gene Homologous to SPTREMBL-ID:Q95368 HLA CLASS I INHIBITORY NK RECEPTOR - HOMO SAPIENS (HUMAN), 4\$5 aa.	Human Gene Similar to SPTREMBL-ID:P91197 SIMILAR TO LIGAND-GATED IONIC CHANNEL PROTEIN - CAENORHABDITIS ELEGANS, 461 aa.
kinesin	МНС	MHC	мнс	misc_channel
SILENT- NONCOD ING	SILENT- NONCOD ING	SILENT- NONCOD ING	SILENT. NONCOD ING	SILENT- NONCOD ING
U	<u>ن</u>	U	U	ල ල
<	∢	.	[
TCAGGAGCAAGGC / GAATGTATGACA[A /C]CATGTCCACAAT GGTGTACATAAAG	TTCTGAAGAGGCT GACGATTTTACT[A/ GJTCTCATTTTTTC CTTTCTCCAGAA	CTAGCTTCCCTTCC CATTCAACACA[A/ CJACACACATTCTT GCTCTACCCAAAG	TGTCTCAAACCCA GCTTGCCAGCTC[T/ CJAATGTACCAGCA GCTGGAATCTGAA	GTGGCTGGGCTAT TCCATCCATCTG[T/ GJAAGCACATTTGA GCCTCCAGGCTTC
374	305	1167	1122	2506
cg43925424	cg42479188	cg42686658	cg38337333	cg27803682
291	292	293	294	295

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				·
7.90E-79	7.508-79	7.50E-79	2.305-71	· .
Human Gene Similar to SPTREMBL-ID:P91197 SIMILAR TO LIGAND-GATED IONIC CHANNEL PROTEIN - CAENORHABDITIS ELEGANS, 461 aa.	Human Gene Similar to SPTREMBL-ID:P91197 SIMILAR TO LIGAND-GATED IONIC CHANNEL PROTEIN - CAENORHABDITIS ELEGANS, 461 aa.	Human Gene Similar to SPTREMBL-ID:P91197 SIMILAR TO LIGAND-GATED IONIC CHANNEL PROTEIN - CAENORHABDITIS ELEGANS, 461 aa.	Human Gene Similar to SPTREMBL-ID:P91197 SIMILAR TO LIGAND-GATED IONIC CHANNEL PROTEIN - CAENORHABDITIS ELEGANS, 461 aa.	Human Gene SWISSPROT- ID:P15498 VAV PROTO- ONCOGENE - HOMO SAPIENS (HUMAN), 846 aa.
misc_channel	misc_channel	misc_channel	misc_channel	oncogene
SILENT- NONCOD ING	SILENT. NONCOD ING	SILENT. NONCOD ING	SILENT. NONCOD ING	SILENT- NONCOD ING
	U	O	ပ	ပ
<u>L</u>	⊢	∢	4	deg
CGAGCGGCACCCA GAGCCTGCACCGT /GJCCCTCACCGTCC TTCTGCGTCCCCC	AGGAGCCTCTTC GGTGTCCCGAGIT /C]GCCACGGTCAA GACCCGCAGCACC A	CGGTCAAGACCCG CAGCACCAAAGCI A/G]CCGCCCCGC ACCTGCCCCTGTCG C	GAGCCGTGTGGCT GTGGCCTCCGGG[A /C]GGCGGTGGACG GCGTGCGCTTCATC	GGGTGCACGGCCG GCCTGGGCAGG[g ap/C]GTAGCCATGG AGCTGTGGCGCCA AT
1440		1890	1541	88
cg214132 <i>67</i>	cg21413267	cg214132 <i>67</i>	cg42481172	cg39518465
296	297	298	299	300

(22q11)			
(33)		∞	
2.40E-84	1.40E-62	3.90E-62	8.00E-257
Human Gene Similar to SWISSPROT-ID:Q64010 PROTO- ONCOGENE C-CRK (P38) (ADAPTER MOLECULE CRK) - MUS MUSCULUS (MOUSE), 304 aa.	Human Gene Similar to SWISSPROT-ID:P31695 NEUROGENIC LOCUS NOTCH HOMOLOG PROTEIN 4 PRECURSOR (TRANSFORMING PROTEIN INT-3) - MUS MUSCULUS (MOUSE), 1964 aa.	Human Gene Similar to TREMBLNEW-ID:G2952331 ARG/ABL-INTERACTING PROTEIN ARGBP2A - HOMO SAPIENS (HUMAN), 666 aa.	Human Gene SWISSNEW- ID:P08684 CYTOCHROME P450 3A4 (EC 1.14.14.1) (CYPIIJA4) (NIFEDIPINE OXIDASE) (NF-25) (P450-PCN1) - HOMO SAPIENS (HUMAN), 502 aa.pcis:SWISSPROT-ID:P08684 CYTOCHROME P450 IIIA4 (EC 1.14.14.1) (NIFEDIPINE OXIDASE) (NF-25) (P450-PCN1) - HOMO SAPIENS (HUMAN), 502 aa.
oncogene	oncogene	опсовепе	oxidase
SILENT- NONCOD ING	SILENT. NONCOD ING	SILENT- NONCOD ING	SILENT- NONCOD ING
U	Ŧ	U	F
gap	U	∢	₹
ATGGGGCCGGTGT CTCGCCAGGAGG[g ap/C]GCAGACCCGG CTCCAGGGCCAGC GC	AGCATTTGAGGAA GCATAACTGACG[C ATGTGAAGGGGGT GTGGGGTACTTGC C	AGCATCTGCAGAC GACCCCGCAGC[A /CJTTTCCCTCGGAC CCCCTCGAAGCC	CACTGCTGTGCAG GGCAGGGA[A/T]GC TCCAGGCAGACAG CCCAGCAAAG
627	235	2295	53
j	cg40333812		cg44014837
301	302	303	304

3 (3q26.3)	-	01	
0	4.60E-246	2.60E-227	1.90E-202
Human Gene SWISSPROT- ID:P07202 THYROID PEROXIDASE PRECURSOR (EC 1.11.1.8) (TPO) - HOMO SAPIENS (HUMAN), 933 aa.	Human Gene SPTREMBL- ID:Q15172 PROTEIN PHOSPHATASE 2A B56-ALPHA - HOMO SAPIENS (HUMAN), 486 aa.	Human Gene SWISSPROT- D:Q14642 TYPE I INOSITOL- 1,4,5-TRISPHOSPHATE 5- PHOSPHATASE (EC 3.1.3.56) (5PTASE) - HOMO SAPIENS (HUMAN), 412 aalpcis:SPTREMBL-ID:Q14642 INOSITOL 1,4,5-TRIPHOPHATE 5-PHOSPHATASE - HOMO SAPIENS (HUMAN), 412 aa.	Human Gene SWISSPROT- ID:P36876 PROTEIN PHOSPHATASE PP2A, 55 KD REGULATORY SUBUNIT, ALPHA ISOFORM (PROTEIN PHOSPHATASE PP2A B SUBUNIT ALPHA ISOFORM) (ALPHA-PR55) - RATTUS NORVEGICUS (RAT), 447 aa.
peroxidase	phosphatase	phosphatase	phosphatase
SILENT- NONCOD ING	SILENT. NONCOD ING	SILENT- NONCOD ING	SILENT. NONCOD ING
			·
gap	O	O	· .
∢	L	4	U
CAGCACAGCGAGC GCTCTCATTCTG[A/ gap]CCTTTTTTCCTC TTCTCAGCCAACT	TCTGTAGAGCTCTG AAAAGGTTGAC[I/ GJATATAGAGGTCT TGTATGTTTTTAC	GACAGACGAGACA GTGAGGTATGTG[A /G]GGCTGCTCCGG AATGGTCCGGAGG C	TAACTATGCAAGA CAAGACTTGGTC[C /GJTCACGTTCGCGT CTCTAGTTGATTT
3178	2958	1537	581
cg41626506	cg43918944	cg43988365	cg43969460
305	306	307	308

2 (2p23)	o	1 (1p21)
1.60E-181	1.20E-89	5.40E-284
Human Gene SWISSPROT- ID:P37140 SERINE/THREONINE PROTEIN PHOSPHATASE PPI- BETA CATALYTIC SUBUNIT (EC 3.1.3.16) (PP-1B) - HOMO SAPIENS (HUMAN), RATTUS NORVEGICUS (RAT), MUS MUSCULUS (MOUSE),, 327 aa.	Human Gene Similar to SWISSPROT-ID:P39687 POTENT HEAT-STABLE PROTEIN PHOSPHATASE 24 INHIBITOR 11PP2A (HLA-DR ASSOCIATED PROTEIN I) (PHAPI) (ACIDIC NUCLEAR PHOSPHOPROTEIN PP32) (CEREBELLAR LEUCINE RICH ACIDIC NUCLEAR PROTEIN) - HOMO SAPIENS (HUMAN), 249 aa.	Human Gene SWISSPROT- ID:P22001 VOLTAGE-GATED POTASSIUM CHANNEL PROTEIN KV1.3 (HPCN3) (HGK5) (HUKIII) (HLK3) - HOMO SAPIENS (HUMAN), 523 aa.
phosphatase	phosphataseinhi b	potassium_chan nel
SILENT- NONCOD ING	SILENT- NONCOD ING	SILENT. NONCOD ING
	C)	D
AATTAAAACTCTA GGTGTATACTTA[I/ CJATGGAACTAGTT TATTTCCTATTTA	TGCTCGCGCGTG CCACTAAGGTCA[C /I]TCCCGCCTCCGA GAGCCCAGAGCCG	CTTTCCCTCTAC (CTCTCT[G/I]) ACCTCTCTCT[G/I] AACATCGTAAACA ACAGACTTACGT
362	215	1977
)		cg42937321
309	310	311

1 (Jp21)	(11q24)
5.40E-284	1.80E-205
SILENT- potassium_chan Human Gene SWISSPROT- NONCOD nel ID:P22001 VOLTAGE-GATED ING POTASSIUM CHANNEL PROTEIN KVI.3 (HPCN3) (HGK5) (HUKII) (HLK3) - HOMO SAPIENS (HUMAN), 523 aa.	Human Gene SWISSPROT- ID:P48048 ATP-SENSITIVE INWARD RECTIFIER POTASSIUM CHANNEL, IWWARDLY RECTIFYING, SUBFAMILY I, MEMBER 1) (ATP-REGULATED POTASSIUM CHANNEL ROM- K) (KIR1.1) - HOMO SAPIENS (HUMAN), 391 aa.
potassium_chan nel	SILENT- potassium_chan NONCOD nel ING
SILENT- pot NONCOD nel ING	SILENT. NONCOD ING
<u> </u>	<u> </u>
U	¥ .
CCTCTTACCCTCTC C TCTCTGAACAT[C/T] JGTAAACAACAGA CTTACGTTAAACT	CAAAATGTAACAG / TGGCTTTTCAAC[A GJGGAGTAAAGCA AAGTCTCTAAAGC T
	1357
cg42937321 1983	cg40991963
312	313

(1925.2)
Human Gene SWISSNEW- ID:P35354 PROSTAGLANDIN GH SYNTHASE 2 PRECURSOR (EC 1.14.99.1) (CYCLOOXYGENASE -2) (COX- 2) (PROSTAGLANDIN- ENDOPEROXIDE SYNTHASE 2) (PROSTAGLANDIN H2 SYNTHASE 2) (PGH SYNTHASE 2) (PGHS-2) (PHS II) - HOMO SAPIENS (HUMAN), 604 aa.lpcls:SPTREMBL-ID:Q16876 PROSTAGLANDIN- ENDOPEROXIDE SYNTHASE-2 PRECURSOR (EC 1.14.99.1) (PROSTAGLANDIN- ENDOPEROXIDE SYNTHASE) (PROSTAGLANDIN- SYNTHASE) (PROSTAGLANDIN SYNTHASE) (PROSTAGLANDIN SYNTHASE) (PROSTAGLANDIN SYNTHASE) (PROSTAGLANDIN SYNTHASE) (HUMAN), 604 aa.
SILENT- prostaglandin NONCOD ING
SILENT- NONCOD ING
∢
AAAGATGTTTGAA G TACTTAAACACT[G/ AJTCACAAGATGGC AAAATGCTGAAAG
cg43951366 2332
314

(1q25.2)	1 (1p31.2)
0	1.40E-211
Human Gene SWISSNEW- ID:P35354 PROSTAGLANDIN G/H SYNTHASE 2 PRECURSOR (EC 1.14.99.1) (CYCLOOXYGENASE -2) (COX- 2) (PROSTAGLANDIN- ENDOPEROXIDE SYNTHASE 2) (EROSTAGLANDIN H2 SYNTHASE 2) (PGH SYNTHASE 2) (PGHS-2) (PHS II) - HOMO SAPIENS (HUMAN), 604 aa. pcls:SPTREMBL-ID:Q16876 PROSTAGLANDIN ENDOPEROXIDE SYNTHASE) (PROSTAGLANDIN ENDOPEROXIDE SYNTHASE) (PROSTAGLANDIN ENDOPEROXIDE SYNTHASE) (PROSTAGLANDIN SYNTHASE)	Human Gene SPTREMBL- ID:000325 PROSTAGLANDIN EP3 RECEPTOR SUBTYPE ISOFORM - HOMO SAPIENS (HUMAN), 402 aa.
prostaglandin	prostaglandin
SILENT- NONCOD ING	SILENT: NONCOD ING
U	O
	i i
TGGTGGAGCCACT GCAGTGTTATCT[T/ C]AAAATAAGAAT ATTTGTTGAGATA	CACTTAACTTGCAT GTGCACAGCTT[IVC JTGGTAACAAATAT CGCTAAACCTTA
2829	1431
10	cg43306254
315	316

1 (1p31.2)	((11q21)	2 (2q13)
1.40E-211	2.40E-146	2,40E-82
Human Gene SPTREMBL- D:000325 PROSTAGLANDIN EP3 RECEPTOR SUBTYPE ISOFORM - HOMO SAPIENS (HUMAN), 402 aa.	Human Gene Homologous to SWISSNEW-ID:P09237 MATRILYSIN PRECURSOR (EC 34.24.23) (PUMP-1 PROTEASE) (UTERINE METALLOPROTEINASE-7) (MATRIX METALLOPROTEINASE-7) (MAMP-7) (MATRIN) - HOMO SAPIENS (HUMAN), 267 aa. pcls:SWISSPROT-ID:P09237 MATRILYSIN PRECURSOR (EC 34.24.23) (PUMP-1 PROTEASE) (UTERINE METALLOPROTEINASE) (MATRIX METALLOPROTEINASE-7) (MMP-7) (MATRIN) - HOMO SAPIENS (HUMAN), 267 aa.	Human Gene Similar to SWISSPROT-ID:P25155 COAGULATION FACTOR X PRECURSOR (EC 3.4.21.6) (STUART FACTOR) (VIRUS ACTIVATING PROTEASE) (VAP) - GALLUS GALLUS (CHICKEN), 475 aa.
prostaglandin	protease	protease
SILENT- NONCOD ING	SILENT- NONCOD ING	SILENT: NONCOD ING
<u></u>	:	C .
ATGTGATTAATTAT A GTGATGAAAAC[A/ TJTTTTTATAAAT GATCTTGGTCTAT	CAATCAGAATTGA O TAAGCACTGTTC[C/ TJTCCACTCCATTT AGCAATTATGTCA	TCCATCCCTCTTTT G GGGCTCTTCTG[G/C JAGGGAAGTAACA TTTACTGAGCACC
1666	1064	1703
	cg42918089	cg44032168
317	318	319

11 (11q22)	6 (6 pter)	6 (6 pter)
2.405-59	1.60E-124	1.60E-124
Hunan Gene Similar to SWISSPROT-ID:P50280 MATRILYSIN PRECURSOR (EC 3.4.24.23) (PUMP-1 PROTEASE) (UTERINE METALLOPROTEINASE) (MATRIX METALLOPROTEINASE-7) (MMP-7) (MATRIN) - RATTUS NORVEGICUS (RAT), 267 aa.	Human Gene Homologous to SWISSPROT-ID-P16083 NAD(P)H DEHYDROGENASE (QUINONE) Z (EC 1.6.99.2) (QUINONE REDUCTASE) (DT- DIAPHORASE) (AZOREDUCTASE) (PHYLLOQUINONE REDUCTASE) + HOMO SAPIENS (HUMAN), 231 aa.	Human Gene Homologous to SWISSPROT-ID-P16083 NAD(P)H DEHYDROGENASE (QUINONE) 2 (EC 1.6.99.2) (QUINONE REDUCTASE) (DT- DIAPHORASE) (AZOREDUCTASE) (PHYLLOQUINONE REDUCTASE) (MENADIONE REDUCTASE) - HOMO SAPIENS (HUMAN), 231 aa.
protease	reductase	reductase
SILENT- NONCOD ING	SILENT- NONCOD ING	SILENT. NONCOD ING
		i.i
ပ	U .	Ð
T	• · · · · · · · · · · · · · · · · · · ·	dg8
TACCCGGAAGTTG AGCTCAATTTCA[T/ CITTCTGTTTTCTGG CCACAACTGCCA	CCCAGTCCTGCGG CTCCTACTGGGGIA CGGTGCGCTGGTC GGAAGATTGCTGG A	TACTGGGGAGTGC GCTGGTCGGAAG[g up/GJATTGCTGGAC TCGCTGAAGAGAG AC
1250	10	161
		cg43927549
320	321	322

6 (6pter)	2	en.	11
1.60E-124	3.30E-207	1.70E-200	2.10E-124
Human Gene Homologous to SWISSPROT-ID-P16083 NAD(P)H DEHYDROGENASE (QUINONE) 2 (EC 1.6.99.2) (QUINONE REDUCTASE) (DT- DIAPHORASE) (AZOREDUCTASE) (PHYLLOQUINONE REDUCTASE) - HOMO SAPIENS (HUMAN), 231 aa.	Human Gene SWISSPROT- ID:015142 ACTIN-LIKE PROTEIN 2 - HOMO SAPIENS (HUMAN), 394 aa.	Human Gene SWISSPROT- ID:Q14012 CALCIUM/CALMODULIN- DEPENDENT PROTEIN KINASE TYPE I (EC 2.7.1.123) (CAM KINASE I) - HOMO SAPIENS (HUMAN), 370 aa.	Human Gene Homologous to SPTREMBL-ID:000379 DELTA- CATENIN - HOMO SAPIENS (HUMAN), 792 aa.
reductase	struct	struct	struct
SILENT - NONCOD ING	SILENT- NONCOD ING	SILENT. NONCOD ING	SILENT. NONCOD ING
O		gap	
O deab	V	ပ	A .
CGGTCCGTGGTCC CCGGGGGCGCAG[g ap/G]TCGCAGCGCT CCCGCCCTCCAGG CG	TTCTCAAAAGGCT GGGGGTATTTAT[A /GJTAAGAACTTATT CCAAAGTGACTCT	AGGAAAGCCGGAG AATTGGGGCACG[C /gap]AAGAGGGGG GCTTTGATGACC GC	AGATTCATCAGAA TAGGATTTTGC[A/ C]AAATCCCACCA TATGCTGTTGAGC
52	780	113	1926
cg43927549	cg43947066	cg43923264	cg43942332
323	324	325	326

		
13	7 (7q32)	17
4.80E-110	3.50E-74	1.205-55
Human Gene Homologous to SPTREMBL-ID:Q28910 MUCIN - BOS TAURUS (BOVINE), 600 aa (fragment).	Human Gene Similar to SWISSNEW-ID:P12273 PROLACTIN-INDUCIBLE PROTEIN PRECURSOR (SECRETORY ACTIN-BINDING PROTEIN) (SABP) (GROSS CYSTIC DISEASE FLUID PROTEIN 15) (GCDFP-15) (GP17) - HOMO SAPIENS (HUMAN), 146 aa.pcis:SWISSPROT-ID:P12273 PROLACTIN-INDUCIBLE PROTEIN PRECURSOR (SECRETORY ACTIN-BINDING PROTEIN) (SABP) (GROSS CYSTIC DISEASE FLUID PROTEIN) (SABP) (GROSS CYSTIC DISEASE FLUID PROTEIN 15) (GCDFP-15) - HOMO SAPIENS (HUMAN), 146 aa.	Human Gene Similar to SWISSPROT-ID-P19065 SYNAPTOBREVIN 2 (VESICLE ASSOCIATED MEMBRANE PROTEIN 2) (VAMF-2) - HOMO SAPIENS (HUMAN), AND BOS TAURUS (BOVINE), 115 ag.
struct	struct	Struct
SILENT- NONCOD ING	SILENT- NONCOD ING	SILENT- NONCOD ING
		6 -
5	U	· ·
CCGCTGTCTCTGTC TTCGCTTTTTA[G/I] TCAAGAAGAATAA TGCGACGAAAAT	CCACTTCTGGGA CACATTGCCTT[C/I JIGITTTCTCCAGC ATGCGCTTGCTC	CATCATCATCATA GTTTACTTCAGCIA/ TJCTTAAATCCCCG AGGAGTCTGCCCT
580	146	546
j l	cg42207316	c <u>e</u> 43927885
327	328	329

	· · · · · · · · · · · · · · · · · · ·	
12 (12q24.2)	∞	∞
0	9.80E-269	9.20E-83
Human Gene SWISSPROT- ID:P29475 NITRIC-OXIDE SYNTHASE, BRAIN (EC 1.14.13.39) (NOS, TYPE I) (NEURONAL NOS) (NNOS) - HOMO SAPIENS (HUMAN), 1434 aa.	Human Gene SWISSPROT- ID:P48651 PHOSPHATIDYLSERINE SYNTHASE I (SERINE- EXCHANGE ENZYME I) (EC 2.7.8) (KIAA0024) - HOMO SAPIENS (HUMAN), 473 aa.	Human Gene Similar to SPTREMBL-ID:Q42761 SQUALENE SYNTHASE (EC 2.5.1.21) (FARNESYL- DIPHOSPHATE FARNESYLTRANSFERASE) (FARNESYLTRANSFERASE) (PRESQUALENE-DI- DIPHOSPHOSPHATE SYNTHASE) - GLYCYRRHIZA GLABRA, 412 aa.
synthase .	synthase	synthase
SILENT- NONCOD ING	SILENT- NONCOD ING	SILENT- NONCOD ING
E-	F	O
gap	O	<u></u>
CTCTTGCCCAGCCG gap GCTGCAAGTTT[gap /TjGTAAGCGCGGG ACAGACACTGCTG A	AGGITACCAAACA GGAATACAACACIC //ITCTCTCCCTTTT CTGCTCTAGAAGG	TGGGTGATGATCA CTGTGCTGCTTG[T/ CJGGCTCATGGCAG AGCATTCAGTGCC
	555	1565
cg40388639 5029	cg43949316	cg43958714
330	331	332

1 (1q23)	1 (1q23)
3.20E-65	3.20E-65
Human Gene Similar to SWISSPROT-ID:P70490 MILK FAT GLOBULE-EGF FACTOR 8 PRECURSOR (MFG-E8) (O- ACETYL GD3 GANGLIOSIDE SYNTHASE) (AGS) (MFGM)- RATTUS NORVEGICUS (RAT), 427 aa, pcls:SFTREMBL- ID:P70490 O-ACETYL GD3 GANGLIOSIDE SYNTHASE - RATTUS NORVEGICUS (RAT), 427 aa.	Human Gene Similar to SWISSPROT-ID-P70490 MILK FAT GLOBULE-EGF FACTOR 8 PRECURSOR (MFG-E8) (O- ACETYL GD3 GANGLIOSIDE SYNTHASE) (AGS) (MFGM) - RATTUS NORVEGICUS (RAT), 427 aa, pcls::SFTREMBL- ID:P70490 O-ACETYL GD3 GANGLIOSIDE SYNTHASE - RATTUS NORVEGICUS (RAT), 427 aa.
synthase	synthase
SILENT- NONCOD ING	SILENT- NONCOD ING
E	O
U	F
ACAGACTGGCTGC C AGCATTAGGAAT[C //JAGGTCATTCCGA AACTCATCATTGA	GGTCATTCCGAAA CTCATGAA[T/ CJCAGGAAGAAGA AGAGTTCAATCTT A
cg43275028 2508	cg43275028
333	334

1 (1923)	1 (1923)
3.20E-65	3.20E-65
Human Gene Similar to SWISSPROT-ID:P70490 MILK FAT GLOBULE-EGF FACTOR 8 PRECURSOR (MFG-E8) (O- ACETYL GD3 GANGLIOSIDE SYNTHASE) (AGS) (MFGM) - RATTUS NORVEGICUS (RAT), 427 aa. pcls:SPTREMBL- ID:P70490 O-ACETYL GD3 GANGLIOSIDE SYNTHASE - RATTUS NORVEGICUS (RAT), 427 aa.	Human Gene Similar to SWISSPROT-ID:P70490 MILK FAT GLOBULE-EGF FACTOR 8 PRECURSOR (MFG-E8) (O- ACETYL GD3 GANGLIOSIDE SYNTHASE) (AGS) (MFGM) - RATTUS NORVEGICUS (RAT), 427 aa.[pcls:SFTREMBL- D:P70490 O-ACETYL GD3 GANGLIOSIDE SYNTHASE - RATTUS NORVEGICUS (RAT), 427 aa.
synthas e	synthase
SILENT- NONCOD ING	SILENT- NONCOD ING
0	O
4	4
AGAATGGCACTGA / ATTCGTTTCTC(A/ GJAACACAGATATA ATTGTTGGTTCAA	CTTTCACFTGGTGC / TGGAGAATTCA[A/ GJAAGTCAAGAAC ATGCTAAGCATAA G
	2873
cg43275028 2601	cg43275028
	. 336

	1 (1923)
3.20E-65	3.20E-65
Human Gene Similar to SWISSPROT-ID:P70490 MILK FAT GLOBULE-EGF FACTOR 8 PRECURSOR (MFG-E8) (O- ACETYL GD3 GANGLIOSIDE SYNTHASE) (AGS) (MFGM) - RATTUS NORVEGICUS (RAT), 427 aa, ptols:SPTREMBL- ID:P70490 O-ACETYL GD3 GANGLIOSIDE SYNTHASE - RATTUS NORVEGICUS (RAT), 427 aa.	Human Gene Similar to SWISSPROT-ID:P70490 MILK FAT GLOBULE-EGF FACTOR 8 PRECURSOR (MFG-E8) (O- ACETYL GD3 GANGLIOSIDE SYNTHASE) (AG3) (MFGM) - RATTUS NORVEGICUS (RAT), 427 aa, pleis:SPTREMBL- ID:P70490 O-ACETYL GD3 GANGLIOSIDE SYNTHASE - RATTUS NORVEGICUS (RAT), 427 aa.
synthase	synthase
SILENT- NONCOD ING	SILENT- NONCOD ING
Α	Ψ.
TTCAAAAGTCAAG AACATGCTAAGC[A /GJTAAGGGACCCA AGGTAGAAAGAGA T	TTCTCCTTCCAGAA TGAGGCCTGG[A G]AGGACCCTCCTA GTGATCTGTTACT
	3073
cg43275028 2894	cg43275028
337	338

1 (1q23)	4	17	1 (1p31.2)
3.20E-65	1.60E-236	2.00E-220	4.80E-212
Human Gene Similar to SWISSPROT-ID:P70490 MILK FAT GLOBULE-EGF FACTOR 8 PRECURSOR (MFG-E8) (O- ACETYL GD3 GANGLIOSIDE SYNTHASE) (AGS) (MFGM) - RATTUS NORVEGICUS (RAT), 427 aa. pick:SPTREMBL- ID:P70490 O-ACETYL GD3 GANGLIOSIDE SYNTHASE - RATTUS NORVEGICUS (RAT), 427 aa.	Human Gene SWISSPROT- ID:P25101 ENDOTHELIN-1 RECEPTOR PRECURSOR (ET-A) - HOMO SAPIENS (HUMAN), 427 aa.	Human Gene SWISSPROT- ID:P51575 P2X PURINOCEPTOR 1 (ATP RECEPTOR) (P2X1) (PURINERGIC RECEPTOR) - HOMO SAPIENS (HUMAN), 399 aa.	Human Gene SWISSPROT- D.P43115 PROSTAGLANDIN E2 RECEPTOR, EP3 SUBTYPE (PROSTANOID EP3 RECEPTOR) (PGE RECEPTOR, EP3 SUBTYPE) - HOMO SAPIENS (HUMAN), 390 aa.
synthase	tm7	TEI .	7 .
SILENT- NONCOD ING	SILENT. NONCOD ING	SILENT- NONCOD ING	SILENT- NONCOD ING
<u>0</u>	0	0	O
∀	U	ပ	· [==
ACTACATAAGGAC AGCAACATGCCT[A /G]TGGACATGAGA GAATTTGTCTTACT	GAAAAAATCACA AGGCAACTGTGA[C /GJTCCGGGAATCT CTTCTCTGATCCTT		ATAATCCATGCCTC TGAATATTAGA[T/ GJTGGTTTCTTGGA TGGGATTTTGAAT
2590	1856	1684	1603
cg43275028	cg43985000	cg39565524	cg43306266
	340	341	342

1 (1p31.2)	1 (1p31.2)
4.80E-212	4.80E-212
Human Gene SWISSPROT- ID:P43115 PROSTAGLANDIN E2 RECEPTOR, EP3 SUBTYPE (PROSTANOID EP3 RECEPTOR) (PGE RECEPTOR, EP3 SUBTYPE) - HOMO SAPIENS (HUMAN), 390 aa.	Human Gene SWISSPROT- ID:P43115 PROSTAGLANDIN E2 RECEPTOR, EP3 SUBTYPE (PROSTANOID EP3 RECEPTOR) (PGE RECEPTOR, EP3 SUBTYPE) - HOMO SAPIENS (HUMAN), 390 aa.
fm7	rm7
SILENT- tm7 NONCOD ING	SILENT- Im7 NONCOD ING
O	ပ
gap	_ප
343 cg43306266 1641 GGGATTTTGAATA gap TGCATTTAAGAA[g ap/C]GTTGGGAAGA ATTTCACAGATGA TG	GAATATGCATTTA G AGAAGTTGGGAA[G/C]AATTTCACAG ATGATTGGAG GA
1641	
cg43306266	cg43306266 1650
343	344

L	*
100	
8.20E-201	2.00E-197
Y12) IN- EG-1) R-+BR) N), (Y12) (f0 OMO a.	Human Gene SWISSPROT- ID:P50052 TYPE-2 ANGIOTENSIN II RECEPTOR (AT2) - HOMO SAPIENS (HUMAN), 363 aa.
tm7	tm <i>7</i>
SILENT. NONCOD ING	SILENT. NONCOD ING
E-	gap
TCGGCAAATCTTG C AAAGCTGCAGGG[C/TJGCAGAGACAT GGATGTGACTTCC CA	AAGGCATAAGAAC TAGGAGCTGCTG[g ap/G]ACATTTCAAT ATGAAGGGCAACT CC
	439
cg43329467	cg2751286
345	346

-			3 (3q21)
3.20E-176	1.10E-173	1.10E-173	6.90E-109
Human Gene SWISSPROT- ID:P46089 PROBABLE G PROTEIN-COUPLED RECEPTOR GPR3 (ACCA ORPHAN RECEPTOR) - HOMO SAPIENS (HUMAN), 330 aa.	Human Gene SWISSPROT- ID:P30542 ADENOSINE A1 RECEPTOR - HOMO SAPIENS (HUMAN), 326 aa.	Human Gene SWISSPROT- ID:P30542 ADENOSINE A1 RECEPTOR - HOMO SAPIENS (HUMAN), 326 aa.	Human Gene Homologous to SWISSPROT-ID:P31421 METABOTROPIC GLUTAMATE RECEPTOR 2 PRECIRSOR - RATTUS NORVEGICUS (RAT), 872 aa.
tm7	tm7	Im.7	tm.7
SILENT- NONCOD ING	SILENT- NONCOD ING	SILENT- NONCOD ING	SILENT. NONCOD ING
	£	U	CD .
U	C)	<i>r</i> n	
GAATGTGGGGATA C AGGCATTGGGAC[C /T]CTATCAGGTATC CTGAGGAGAGACT	CAGCGGGAGCTC TGCCAGCTTTGG[C/ TJGAAGGAGGGTG CTTGCCTCGTGCCC	CGGGAGCTCTGCC AGCTTTGGCGAA[G /C]GAGGGTGCTTG CCTCGTGCCCTTG	TGCTCTTGCTGCTG A ATGGAGGAGGA[A/ GJGGGGTGGATCCC GTGGAGCCTCCAA
9/	135	139	1839
cg11751407	cg43326635	cg43326635	cg43993798
347	348	349	350

2.90E-74	2.90E-74
Human Gene Similar to SWISSPROT-ID:Q25322 TYRAMINE/OCTOPAMINE RECEPTOR 2 (TYR-LOC 2)- LOCUSTA MIGRATORIA (MIGRATORY LOCUST), 484 aa_lpels:SPTREMBL-ID:Q25322 GCR2 (G PROTEIN-COUPLED RECEPTOR) - LOCUSTA MIGRATORY LOCUSTA	Human Gene Similar to SWISSPROT-ID:Q25322 TYRAMINE/OCTOPAMINE RECEPTOR 2 (TYR-LOC 2)- LOCUSTA MIGRATORIA (MIGRATORY LOCUST), 484 aa. lpcls:SPTREMBL-ID:Q25322 GCR2 (G PROTEIN-COUPLED RECEPTOR) - LOCUSTA MIGRATORY (MIGRATORY LOCUST), 484 aa.
tm.7	11.
SILENT- NONCOD ING	SILENT- NONCOD ING
ပ	
<u> </u>	O
ATGCTTCCCCCAAC A CCTAGGGAATC(A/CJACACTTAAGATA ATTCGCCACTTCT	CCAACCCTAGGGA A TCAACACTTAA[G //IJATAATTCGCCAC TTCTCCTTTCT
	2139
cg43040271 2130	cg43040271
351	

	5 (5q32)	<i>S</i> (5q32)	<i>5</i> (5q32)
2.90E-74	2.00E-58	2.00E-58	2.00E-58
Human Gene Similar to SWISSPROT-ID:Q25322 TYRAMINE/OCTOPAMINE RECEPTOR 2 (TYR-LOC 2) - LOCUSTA MIGRATORIA (MIGRATORY LOCUST), 484 aa.lpcis:SPTREMBL-ID:Q25322 GCR2 (G PROTEIN-COUPLED RECEPTOR) - LOCUSTA MIGRATORIA (MIGRATORY LOCUST), 484 aa.	Human Gene Similar to SWISSPROT-ID:Q24563 DOPAMINE RECEPTOR 2 - DROSOPHILA MELANOGASTER (FRUIT FLY), 539 aa.	Human Gene Similar to SWISSPROT-ID:Q24563 DOPAMINE RECEPTOR 2 - DROSOPHILA MELANOGASTER (FRUIT FLY), 539 aa.	Human Gene Similar to SWISSPROT-ID:Q24563 DOPAMINE RECEPTOR 2 - DROSOPHILA MELANOGASTER (FRUIT FLY), 539 aa.
m <i>)</i>	tm7	tm.7	tm7
SILENT. NONCOD ING	SILENT- NONCOD ING	SILENT: NONCOD ING	SILENT- NONCOD ING
E-r	υ ·	D D	Ů
O	<u></u>	∀	ບ
AGATAATTCGCCA CTTCTCCTCTT{C/ TJTCTCTGCTCCGC TCACGGCTTGCAG	CGCAGAGCCCCGC CGTGGGTCCGCCTV CJGCTGAGGCGCCC CCAGCCAGTGCGC	CAGCGCCTTCTTGC TGGCACCCAAT[A/ GJGAAGCCATGCGC CGGACCACGACGT	TGCGCCGGACCAC GACGTCACGCAG[C /G]AAAGGGACGAG GTGTGGGTGGTGG G
	1668	1760	1793
cg43040271	cg43040273	cg43040273	cg43040273
353	354	355	356

5 (5q32)	5 (5 q32)	5 (5q32)	12	12
2.00E-58	2.00E-58	2.00E-58	1.70E-177	1.70E-177
Human Gene Similar to SWISSPROT-ID:Q24563 DOPAMINE RECEPTOR 2 - DROSOPHILA MELANOGASTER (FRUIT FLY), 539 aa.	Human Gene Similar to SWISSPROT-ID:Q24563 DOPAMINE RECEPTOR 2 - DROSOPHILA MELANOGASTER (FRUIT FLY), 539 aa.	Human Gene Similar to SWISSPROT-ID:Q24563 DOPAMINE RECEPTOR 2 - DROSOPHILA MELANOGASTER (FRUIT FLY), 539 aa.	Human Gene SPTREMBL- ID:Q07279 TRANSCRIPTION FACTOR NF-E2 - MUS MUSCULUS (MOUSE), 373 aa.	Human Gene SPTREMBL- ID:Q07279 TRANSCRIPTION FACTOR NF-E2 - MUS MUSCULUS (MOUSE), 373 aa.
tm7	tm7	tm7	transcriptfactor	transcriptfactor
SILENT- NONCOD ING	SILENT- NONCOD ING	SILENT. NONCOD ING	SILENT- NONCOD ING	SILENT: NONCOD ING
O	4	A	[mq	U
O	Ð	deg	gap	් ප
GCAGGTCTTCTTTG AAGGCCTATGG[6/ CJAATGGCTACTCC AGCAACGGCAACA	ATTGTAGTACAAA TGACTCACTGCT[G/ AJTAAAGCAGTTTT TCTACTTTTAAAG	ATAAACTTAGAAT AAAATTGTAAAA[g ap/A]TTGTATAGAG ATATGCAGAAGGA AĞ	AGGGGTGGAACTG CTGATGGGATTT[ga p/T]CCTTCATTCCC TTCTGATAAAGGT A	AGCCTCCCAGAG ACAACACCGGGA[GACJCCTCATCTCTC TCCTCACCCTGCTG
2767	2953	3053	1501	249
	cg43040273	cg43040273	cg43998970	cg43998970
357	358	359	360	361

		\$ (\$p15.3)	
4.20E-158 8	6.90E-68	چ د د	0
Human Gene SWISSNEW- ID:P23193 TRANSCRIPTION ELONGATION FACTOR S-II (TRANSCRIPTION ELONGATION FACTOR A) - HOMO SAPIENS (HUMAN), 301 aa. pcis:SWISSPROT-ID:P23193 TRANSCRIPTION ELONGATION FACTOR S-II (TRANSCRIPTION ELONGATION FACTOR S-II (TRANSCRIPTION HOMO SAPIENS (HUMAN), 301 aa.	Human Gene Similar to TREMBLNEW-ID:G2920821 TRANSCRIPTION FACTOR T- BOX 5 - HOMO SAPIENS (HUMAN), 518 aa.	номо	Human Gene SWISSPROT- ACC:P02545 LAMIN A (70 KD LAMIN) - Homo sapiens (Human), 664 aa.
transcriptfactor	transcriptfactor	transport	UNCLASSIFIE D
SILENT. NONCOD ING	SILENT. NONCOD ING	SILENT: NONCOD ING	SILENT- NONCOD ING
E	<u>ප</u>	ن ب	₹
GTCTTCTCCGCGCC C CACCCCGCTGG[CT] JAAGGGGAAGTGG GCGAAGCTGGAGC	GGGCCGGGCACT A GCCCAGGAAGGG[A/G]CTCCGGGAGA GGGAGCCGGCGGC TG	AGACGAAGACCCC T AGGAAGTCATCCTT /CJGCAATGGGAGA GACACGAACAAAC C	CCCACGCCTGCCA gap GGAGCAAGCCGA[g ap/A]GAGCCAGCCG GCCGGCGCACTCC GA
2623	934	2030	237
			cg43921289
362	363	364	365

16	16	91	1 (ip32)	17 (17q11)
0	0	0	0	0
Human Gene SWISSPROT- ACC:Q14687 HYPOTHETICAL PROTEIN KIAA0182 - Homo sapiens (Human), 1157 aa (fragment).	Human Gene SPTREMBL- ACC:Q16084 P130 - HOMO SAPIENS (HUMAN), 1139 aa.	Human Gene SPTREMBL- ACC:Q16084 P130 - HOMO SAPIENS (HUMAN), 1139 aa.	Human Gene SWISSPROT- ACC:P42566 EPIDERMAL GROWTH FACTOR RECEPTOR SUBSTRATE SUBSTRATE 15 (PROTEIN EPS15) (AF-1P PROTEIN) - Homo sapiens (Human), 896 aa.	Human Gene SWISSPROT- ACC.P53675 CLATHRIN HEAVY CHAIN 2 (CLH-22) - Homo sapiens (Human), 1640 aa.
UNCLASSIFIE D	UNCLASSIFIE D	UNCLASSIFIE D	UNCLASSIFIE D	UNCLASSIFIE D
SILENT. NONCOD ING	SILENT: NONCOD ING	SILENT: NONCOD ING	SILENT- NONCOD ING	SILENT. NONCOD ING
O	5	5	U	Ð
∀	U	⋖	₹	4
AAACAAATAAGCC CITITTACTGAC[A/ G]ATGCACCCAACC TTTTCAGCTGAAG	AGAGTCAAAAATC CAAGTTTGGATT[C/ G]TAAGCAGCCTTG ACAGTAATCACTG	AAGCAGCCTTGAC AGTAATCACTGA[A /GJTGGTAGGGAAA AAAAGACAGTTGG G	AGGCAAAAGCTCA CAGTAAATGTAT[A /C]CCAGAACAGGG GCCTAAGTGAAGG T	CTGCTCCCANCTTC GCCAGCCTCCA[A/ G]TGTACAACTTCC GCGTGTAGTGGGC
3196	1309	1336	2206	4893
	cg43955093	cg43955093	cg43925474	cg44014437
366	367	368	369	370

17 (17q11)	20 (20pter)	21 (21q22.1)	22 (22q13.1)
0	0		0
Human Gene SWISSPROT- ACC:P53675 CLATHRIN HEAVY CHAIN 2 (CLH-22) - Homo sapiens (Human), 1640 aa.	Human Gene SWISSPROT- ACC:P05060 SECRETOGRANIN I PRECURSOR (SGI) (CHROMOGRANIN B) - Homo sapiens (Human), 677 aa.	Human Gene SWISSNEW- ACC:Q13009 T-LYMPHOMA INVASION AND METASTASIS INDUCING PROTEIN I (TIAMI PROTEIN) - Homo sapiens (Human), 1591 aa.	Human Gene SWISSPROT- ACC:P13866 SODIUM/GLUCOSE COTRANSPORTER 1 (NA(+)/GLUCOSE COTRANSPORTER 1) (HIGH AFFINITY SODIUM-GLUCOSE COTRANSPORTER) - Homo sapiens (Human), 664 aa.
NCLASSIFIE	UNCLASSIFIE D	UNCLASSIFIE D	UNCLASSIFIE D
SILENT. U NONCOD D ING	SILENT. NONCOD ING	SILENT- NONCOD ING	SILENT- NONCOD ING
5	∢	Į-	Į-
A	U	O	U
CTGCTCCCAACTTC GCCAGCCTCCA[A/ GJTGTACAACTTCC GCGTGTAGTGGGC	CACTTCACTGAAA GACACCATTTAT[C/ AJTACCCAAGGGCA GAAAGTAGAACTT	GATAGGACTCAAG CTTATTGGGAT[C/ TJCTGATGATCT TTCTGATGTTGTT	TACAGCCATCTGT ACCTACTGGAGG[C /T]GCAGAAGGGAA GTCCACTCAGTCA C
5114	2242	1939	2416
cg44014448	cg43973129	cg43950657	cg43956384
371	372	373	374

23)				
X (Xp22.3)				_
0		0	1.60E-292	1.20E-280
Human Gene SWISSPROT- ACC:P23352 KALLMANN SYNDROME PROTEIN PRECURSOR (ADHESION MOLECULE-LIKE X-LINKED) - Homo sapiens (Human), 680 aa.	Human Gene TREMBLNEW- ACC:AAD23581 CULLIN 2 - HOMO SAPIENS (HUMAN), 745 aa.	Human Gene TREMBLNEW- ACC:AAD23581 CULLIN 2 - HOMO SAPIENS (HUMAN), 745 aa.	Human Gene TREMBLNEW- ACC:CAA08974 GUANINE NUCLEOTIDE-EXCHANGE FACTOR - HOMO SAPIENS (HUMAN), 548 aa.	Human Gene SWISSPROT- ACC:P38567 HYALURONIDASE PRECURSOR (EC 3.2.1.35) (SPERM SURFACE PROTEIN PH-20) (SPERM ADHESION MOLECULE 1) - Homo sapiens (Human), 509 aa.
UNCLASSIFIE D	UNCLASSIFIE D	UNCLASSIFIE D	UNCLASSIFIE D	UNCLASSIFIE D
SILENT- NONCOD ING	SILENT- NONCOD ING	SILENT- NONCOD ING	SILENT: NONCOD ING	SILENT- NONCOD ING
≺	<u> </u>	E	F	V
5	V		<	(←
AGCAGTGCAGCCC CGGCGCGGAGCA[G/A]GGAGCCTCGG CCCGCGCCCGGCG	GAGAAAAGCATG GTACCCAACCGA[A /IJITTCCACTITTC AGCAATACTTCAC	TAAAGTTTTAAGA AATGTCATAATGIA TJCATGAGCTTGA AATATCTCTAGGC A	AGCAAAGAAACAC TGGCAGAATTCCIA TJGCATTTGCAAA ATTCTAAGTTTTGG	AAATAAATGTTTTC ATAGTCATTAC[T/A]CTTTACAATGGGA GTGCTAAAATTC
101	260	323	1121	366
	cg44932392	cg44932392	cg43981656	cg44910613
375	376	377	378	379

Х (Хр22.1)	7	2 (2p25)	
4.70E-261	2.10E-258	7.00E-251	4.80E-213
	Human Gene SPTREMBL- ACC:P78506 DIABETES MELLITUS TYPE I AUTOANTIGEN (ISLET CELL AUTOANTIGEN P69) - HOMO SAPIENS (HUMAN), 483 aa.	Human Gene SWISSPROT- ACC:P11926 ORNITHINE DECARBOXYLASE (EC 4.1.1.17) (ODC) - Homo sapiens (Human), 461 aa.	Human Gene SWISSPROT- ACC:P32754 4- HYDROXYPHENYLPYRUVATE DIOXYGENASE (EC 1.13.11.27) (4HPPD) (HPD) - Homo sapiens (Human), 392 aa.
UNCLASSIFIE D	UNCLASSIFIE D	UNCLASSIFIE D	UNCLASSIFIE D
SILENT- NONCOD ING	SILENT- NONCOD ING	SILENT- NONCOD ING	SILENT- NONCOD ING
U	gap	des	H
]E-1	_ව	ပ	∢
AACTGGGTTGCTCT AAGAACTGATGT7/ CJCTAAACCGTCTC AGCATGGCCTGTA	CAATGCATGAATC G TGTACCCTTCGG[G/ gap]AGGGCACTCAC ATGCCGCCCCAG C	TTGTTCATGATTTC TTGATGTTCCT[C/ga p]TAATGGAAAACT AAGAGATGGAATT	GCCGAGTCCGCTG GTGGCGGACCC[A TJAGGGGAGCAGC CAGTAGGGAAGTT G
68	1643	1961	129
cg44035104	cg43929959	cg43950250	cg43064090
380	381	382	383

			4 (4q21.2)	1 0
4.80E-213	4.80E-213	4.80E-213	1.20E-177 (4.20E-166
Human Gene SWISSPROT- ACC:P32754 4- HYDROXYPHENYLPYRUVATE DIOXYGENASE (EC 1.13.11.27) (4HPPD) (HPD) - Homo sapiens (Human), 392 aa.	Human Gene SWISSPROT- ACC:P32754 4- HYDROXYPHENYLPYRUVATE DIOXYGENASE (EC 1.13.11.27) (4HPPD) (HPD) - Homo sapiens (Human), 392 aa.	Human Gene SWISSPROT- ACC:P32754 4- HYDROXYPHENYLPYRUVATE DIOXYGENASE (EC 1.13.11.27) (4HPPD) (HPD) - Homo sapiens (Human), 392 aa.	Human Gene SWISSPROT. ACC:P30968 GONADOTROPIN- RELEASING HORMONE RECEPTOR (GNRH-R) - Homo sapiens (Human), 328 aa.	Human Gene SWISSNEW- ACC:Q16637 SURVIVAL MOTOR NEURON PROTEIN 1 - Homo sapiens (Human), 294 aa.
UNCLASSIFIE D	UNCLASSIFIE D	UNCLASSIFIE D	UNCLASSIFIE D	UNCLASSIFIE D
SILENT- NONCOD ING	SILENT: NONCOD ING	SILENT: NONCOD ING	SILENT. NONCOD ING	SILENT: NONCOD ING
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CCGAGTCCGCTGG / TGGGCGGACCCA[A /T]GGGGAGCAGCC AGTAGGGAAGTTG G	GGGAGCAGT AGGGAAGTTGGG[C/G]GAGTTCCAGA ATCAGGGGGCGTG GC	TAATCGGGAGGGC TGGAGCAGAGGG CGGGCCCCGCCG AGGGGCGTGGTCA GT	GATGCCAAAAAA CAAAGGTGAGAA[A/C]CCACAACACA GGTCTAAACTCAG CA	GTCTTTTACAGATG GTTTTTCAAAA[1/g sp]AGAGTCCAGTA AAATATTTCACATT
130	157	01	3296	381
cg43064090	cg43064090	cg43064090		cg43924431
384	385	386	387	388

13	1 (1p22)	2	9	7
4.30E-164	7.70E-158	4.90E-156	1.10E-150	1.90E-138
Human Gene TREMBLNEW- ACC:AAD40550 P38IP - HOMO SAPIENS (HUMAN), 733 aa.	Human Gene SWISSNEW- ACC.P13726 TISSUB FACTOR PRECURSOR (TF) (COAGULATION FACTOR III) (THROMBOPLASTIN) (CD142 ANTIGEN) - Homo sapiens (Human), 295 aa.	Human Gene SPTREMBL- ACC:Q92600 PROTEIN INVOLVED IN SEXUAL DEVELOPMENT, COMPLETE CDS - HOMO SAPIENS (HUMAN), 299 aa.	Human Gene Homologous to TREMBLNEW-ACC:AAC69899 SACM21 - MUS MUSCULUS (MOUSE), 721 aa.	Human Gene Homologous to TREMBLNEW-ACC:AAD23440 LR8 - HOMO SAPIENS (HUMAN), 270 aa.
UNCLASSIFIE D	UNCLASSIFIE D	UNCLASSIFIE D	UNCLASSIFIE D	UNCLASSIFIE D
SILENT- NONCOD ING	SILENT- NONCOD ING	SILENT- NONCOD ING	SILENT: NONCOD ING	SILENT- NONCOD ING
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CGTTGTTCCTAATG TGGATCTACCA[CT JCCCTGTGTTCATC GAGATTCCGGTC	TGGGATTACAGGT GCGCACTACCACJA /GJCCAAGCTAATTT TTGTATTTTTTAG	CCTTCAGCACCCCT GCAGCGGAAAA[C/ TJAATGAGCCGCCG TAGCCGCCATCCG	AAAAAGCTACAGA AAAGAAATCACTT /CJTGAAAAACACA ATGACTCAGAGGC A	CAGGGACATGCGG G GCACCCGTGGG[G /gap]TCTTTGGCGGC TCACAGGACAATG G
607	1542	2065	176	418
	cg43272443		cg43964140	cg43285114
389	390	391	392	

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-125	-123	817
3.30E-125	2.70E-123	6.90E-118
Human Gene Homologous to SWISSNEW-ACC:P18582 CD81 ANTIGEN (26 KD CELL SURFACE PROTEIN TAPA-1) - Homo sapiens (Human), 236 aa.	Human Gene Homologous to SPIREMBL-ACC:Q15025 MRNA (HA1652) FOR ORF, PARTIAL CDS - HOMO SAPIENS (HUMAN), 296 aa (fragment).	Human Gene Homologous to SWISSPROT-ACC:P22061 ASPARTATE) O- METHYLTRANSFERASE (EC A.1.1.77) (PROTEIN-BETA- ASPARTATE METHYLTRANSFERASE) (PIMT) (PROTEIN L- ISOASPARTYLD-ASPARTYL METHYLTRANSFERASE) (PIMT) (PROTEIN L- ISOASPARTYL D-ASPARTYL METHYLTRANSFERASE) (L- ISOASPARTYL PROTEIN CARBOXYL METHYLTRANSFERASE) - Homo sapiens (Human), 226 aa.
UNCLASSIFIE D	UNCLASSIFIE D	UNCLASSIFIE D
SILENT- NONCOD ING	SILENT- NONCOD ING	SILENT- NONCOD ING
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() (D 4 (D	TAAACAGCTCAGT TCAGGGACTGGT[A //GJTACAAGCTGGC CACCCATCTCAGC C	TTACAGGACATCA CCTGCCATCTTA[I/I A]GGTTTAATTT ACAAATGCCTAGT
370	649	
1	cg44003626 6	cg43917206 2
394	395	396

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∞		21	22 (22q12.1)
2.50E-111	2.90E-110	2.90E-110	1.20E-106
Human Gene Homologous to SPTREMBL-ACC:000559 CANCER ASSOCIATED SURFACE ANTIGEN - HOMO SAPIENS (HUMAN), 213 aa.	Human Gene Homologous to SPTREMBL-ACC:P97314 DOUBLE LIM PROTEIN-1 - MUS MUSCULUS (MOUSE), 193 aa.	Human Gene Homologous to SPTREMBL-ACC:P97314 DOUBLE LIM PROTEIN-1 - MUS MUSCULUS (MOUSE), 193 aa.	Human Gene Homologous to SWISSPROT-ACC:P15018 LEUKEMIA INHIBITORY FACTOR PRECURSOR (LIF) (DIFFERENTIATION- STIMULATING FACTOR) (D FACTOR) (MELANOMA- DERIVED LPL INHIBITOR) (MLPL) - Homo sapiens (Human), 202 ag.
UNCLASSIFIE D	UNCLASSIFIE D	UNCLASSIFIE D	UNCLASSIFIE D
SILENT- NONCOD ING	SILENT. NONCOD ING	SILENT- NONCOD ING	SILENT- NONCOD ING
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U	U	U	Α
GGCCGATTTTTCCA CAATTTAAATC[C/T JCAGTTCACCTGGT ATCCAGCTCCAG	GTTTCCACCTCCCC AGACAGGCATT[C/ TJCGAGTGGGAGGC GGGAGCACGTACC	TTICCACCTCCCCA GACAGGCATTC[C/ TJGAGTGGGAGGC GGGAGCACGTACC G	CTAAACCCAAATG GGGGCTGCTGGC[A TJGACCCCGAGGG TGCCTGGCCAGTC C
215	840	841	
cg43289666	cg43986282	cg43986282	cg432 <i>977</i> 16
397	398	366	400

8 (8q22)			
7.90E-101	1.00E-100	1.60E-100	1.60E-100
Human Gene Homologous to SWISSPROT-ACC:P34741 SYNDECAN-2 PRECURSOR (FIBROGLYCAN) (HEPARAN SULFATE PROTEOGLYCAN CORE PROTEIN) (HSPG) (SYND2) - Homo sapiens (Human), 201 aa.	Human Gene Similar to SPTREMBL-ACC:043399 HD54+INS2 ISOFORM - HOMO SAPIENS (HUMAN), 206 aa.	Human Gene Similar to SWISSNEW-ACC.P11686 PULMONARY SURFACTANT- ASSOCIATED PROTEIN C PRECURSOR (SP-C) (SP5) (PULMONARY SURFACTANT- ASSOCIATED PROTEOLIPID SPL(VAL)) - Homo sapiens (Human), 197 aa.	Human Gene Similar to SWISSNEW-ACC:P11686 PULMONARY SURFACTANT- ASSOCIATED PROTEIN C PRECURSOR (SP-C) (SP5) (PULMONARY SURFACTANT- ASSOCIATED PROTEOLIPID SPL(VAL)) - Homo sapiens (Human), 197 aa.
UNCLASSIFIE D	UNCLASSIFIE D	UNCLASSIFIE D	UNCLASSIFIE D
SILENT- NONCOD ING	SILENT: NONCOD ING	SILENT- NONCOD ING	SILENT: NONCOD ING
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ပ	O	O	
TTTTATCATTAAAG TGCCAGAATGG[C/ TJTCTTTAATGAAA ACAAAAACAAAG	GGAGGGTTGGAGT CACTGACGAATG[C TJGAGCCGGGCCA GGCCCATGCAAAG G	GCCACCTGCCGG GCTGTGGAGGAG[C /gap]GCTCGCGCTG ACCAGGCGCTGGG GC	GCTTCTGCCCACAC CGCAGGGACAA[A GJCCCTGGAGAAAT GGGAGCNTGGGGA
2160	624	883	1124
cg43980312	cg43939240	cg43941552	cg43941552
401	402	403	404

				
12	22	10	10	01
2.10E-100	5.30E-95	3.40E.93	3.40E-93	3.40E-93
Human Gene Similar to SWISSPROT-ACC:P45973 HETEROCHROMATIN PROTEIN 1 HOMOLOG ALPHA (HP1 ALPHA) (ANTIGEN P25) - Homo sapiens (Human), 191 aa.	Human Gene Similar to SWISSPROT-ACC.P30536 PERIPHERAL-TYPE BENZODIAZEPINE RECEPTOR (PBR) (PKBS) (MITOCHONDRIAL BENZODIAZEPINE RECEPTOR) - Homo sapiens (Human), 169 aa.	Human Gene Similar to SWISSPROT-ACC:P36405 ADP- RIBOSYLATION FACTOR-LIKE PROTEIN 3 - Homo sapiens (Human), 182 aa.	Human Gene Similar to SWISSPROT-ACC.P36405 ADP- RIBOSYLATION FACTOR-LIKE PROTEIN 3 - Homo sapiens (Human), 182 aa.	Human Gene Similar to SWISSPROT-ACC:P36405 ADP- RIBOSYLATION FACTOR-LIKE PROTEIN 3 - Homo sapiens (Human), 182 aa.
UNCLASSIFIE D	UNCLASSIFIE D	UNCLASSIFIE D	UNCLASSIFIE D	UNCLASSIFIE D
SILENT- NONCOD ING	SILENT- NONCOD ING	SILENT- NONCOD ING	SILENT. NONCOD ING	SILENT. NONCOD ING
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CATTICICITIGIA CATAATACATICI JACCICCCIGCCIC CICICCITICIA	CAGGGGTCAGCAG AGCTTCAGAGGT[G //]GCCCCACCTGA GCCCCACCCGGG A	CAGAAAGCAGCAA ATTAGTGTTTTTĮC/ AJAGGACCGAATTC GGCTCCCGCAGCT	AAGCAGCAAATTA GTGTTTTCAGG[A/ C]CCGAATTCGGCT CCCGCAGCTCCTG	CTCCCGCAGCTCCT GCATCTCCATT[CT JGTCTAGATTTTAT TTCTTCTTTGCA
914	878	507	511	547
	cg43927693			c <u>g</u> 43951338
405	406	407	408	409

7.20E-91	7.20E-91	7.20E-91	7.20E-91
Human Gene Similar to SWISSNEW-ACC:P01185 VASOPRESSIN-NEUROPHYSIN 2-COPEPTIN PRECURSOR [CONTAINS: ARG- VASOPRESSIN; NEUROPHYSIN 2 (NEUROPHYSIN-II); COPEPTIN] - Homo sapiens (Human), 164 aa.	Human Gene Similar to SWISSNEW-ACC:P01185 VASOPRESSIN-NEUROPHYSIN 2-COPEPTIN PRECURSOR [CONTAINS: ARG- VASOPRESSIN; NEUROPHYSIN 2 (NEUROPHYSIN-II); COPEPTIN] - Homo sapiens (Human), 164 aa.	Human Gene Similar to SWISSNEW-ACC:P01185 VASOPRESSIN-NEUROPHYSIN 2-COPEPTIN PRECURSOR [CONTAINS: ARG- VASOPRESSIN; NEUROPHYSIN 2 (NEUROPHYSIN-II); COPEPTIN] - Homo sapiens (Human), 164 aa.	Human Gene Similar to SWISSNEW-ACC:P01185 VASOPRESSIN-NEUROPHYSIN 2-COPEPTIN: ARG- VASOPRESSIN; NEUROPHYSIN 2 (NEUROPHYSIN-II); COPEPTINI - Homo sapiens (Human), 164 aa.
UNCLASSIFIE D	UNCLASSIFIE D	UNCLASSIFIE D	UNCLASSIFIE D
SILENT- NONCOD ING	SILENT- NONCOD ING	SILENT- NONCOD ING	SILENT- NONCOD ING
E-4	F	[-u	Į
gap de de de	ບ	O	U
CCCGCCCAGCCG ACGCTACTGAG[g ap/T]CCCCGCGCTC GCCCACCGGCGC GC	CCAGCCGACGCC TACTGAGCCCGIC AJGCTCGCCCCACC GGCGCGCTCTTCG	AGCCCGACGCCTA CTGAGCCCGCGGC //ITCGCCCCACCGG CGCGCTCTTCGCG	CGACGCCTACTGA GCCCCGCCGCCCC TJCCCACCGCCGC GCTCTTCGCG(CCG
1234	1240	1242	1246
cg25236776	cg25236776	cg25236776	cg25236776
410	411	412	413

X (Xp11.4)	11 (11p15.2)	7		11
5.00E-83	2.00E-70	1.60E-67	4.30E-66	4.30E-66
Human Gene Similar to REMTREMBL-ACC:E47283 DNA FOR ORF1 AND ORF2 FROM CHROMOSOME X - HOMO SAPIENS (HUMAN), 157 aa.	Human Gene Similar to SWISSNEW-ACC:P01258 CALCITONIN PRECURSOR - Homo sapiens (Human), 141 aa.	Human Gene Similar to TREMBLNEW-ACC:AAD39844 HSPC028 - HOMO SAPIENS (HUMAN), 419 aa.	Human Gene Similar to TREMBLNEW-ACC:AAD29427 MYOMEGALIN - RATTUS NORVEGICUS (RAT), 2324 aa.	Human Gene Similar to TREMBLNEW-ACC:AAD29427 MYOMEGALIN - RATTUS NORVEGICUS (RAT), 2324 aa.
UNCLASSIFIE D	UNCLASSIFIE D	UNCLASSIFIE D	UNCLASSIFIE D	UNCLASSIFIE D
SILENT- NONCOD ING	SILENT- NONCOD ING	SILENT- NONCOD ING	SILENT- NONCOD ING	SILENT: NONCOD ING
A	L	gap	gap	gap
gap	O	L	V	V
GCTACGTTTACTCA CAGCCAGCGAA[ga p/A]CTGACATTAAA ATAACTAACAAAC A	CGCCTCTGATCCA AGCCACCTCCCG[C TJCAGAGAGGTGT CATGGGCTTCCAA A	CTCTGCACAAGGG AAGCCTATCCTA[T/ gap]TTTTTTTTTCCT TTGCGAAAACAGA	AATGCCTCAGATC AGTGACCCAAGG[A/gap]ACCTTCCAG AATGGATGAAATA GAC	ATGCCTCAGATCA GTGACCCAAGGA[Agap]CCTTCCAGA ATGGATGAAATAG ACC
1362	104	356	1119	1120
	cg42748886	cg43969533	cg43976681	cg43976681
414	415	416	417	418

19	9 (9q22)		-
7.30E-66	6.60E-65	5.90E-64	2.00E-54
Human Gene Similar to SPTREMBL-ACC: 000455 TTF-1 INTERACTING PEPTIDE 20 - HOMO SAPIENS (HUMAN), 385 aa (fragment).	Human Gene Similar to SWISSPROT-ACC:P05062 FRUCTOSE-BISPHOSPHATE ALDOLASE B (EC 4.1.2.13) (LIVER-TYPE ALDOLASE) - Homo sapiens (Human), 363 aa.	Human Gene Similar to SPTREMBL-ACC:076087 GAGE-8 - HOMO SAPIENS (HUMAN), 117 aa.	Human Gene Similar to SWISSNEW-ACC:P47992 LYMPHOTACTIN PRECURSOR (CYTOKINE SCM-1) (ATAC) (LYMPHOTAXIN) (SCM-1- ALPHA) - Homo sapiens (Human), 114 aa.
UNCLASSIFIE D	UNCLASSIFIE D	UNCLASSIFIE D	UNCLASSIFIE D
SILENT: U NONCOD D ING	SILENT- NONCOD ING	SILENT: NONCOD ING	SILENT- NONCOD ING
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lu	<u>ව</u>	_ව	U
CCAAGCGGAAGGC CATTTCCCTGC[C/ TJCTTCCTCAGTTG TCCGGGGGGGGG	CTAATTGTGTGGA ATTTCCAGGATT[G/ AJGAGGAAAAGTT GCTCCCTTTCAGCC	AAAGCAATCACAG C TGTTAAAAGAAG[G /A]CACGTTGAAAT GATGCAGGCTGCT C	CCAGCCAGCTCAT TTCACTTTACAC[G/ C]CTCATGGACTGA GTTTATACTCACC
	398	577	423
cg43984044 714	cg43933283	cg42381630	cg41664708
419	420	421	422

13 (13q14.3)	17 (17q21.3 2)	17 (17q21.3 2)	17 (17q21.3 2)
0	0	0	0
ATPase_associat Human Gene SWISSPROT- ed ID:P35670 COPPER- TRANSPORTING ATPASE 2 (EC 3.6.1.36) (COPPER PUMP 2) (WILSON DISEASE- ASSOCIATED PROTEIN) - HOMO SAPIENS (HUMAN), 1465 aa.	Human Gene SWISSPROT- ID:P08514 PLATELET MEMBRANE GLYCOPROTEIN IIB PRECURSOR (GPIIB) (INTEGRIN ALPHA- IIB) (CD41) - HOMO SAPIENS (HUMAN), 1039 aa.	Human Gene SWISSPROT- ID:P08514 PLATELET MEMBRANE GLYCOPROTEIN IIB PRECURSOR (GPIIB) (INTEGRIN ALPHA- IIB) (CD41) - HOMO SAPIENS (HUMAN), 1039 aa.	Human Gene SWISSPROT- ID:P08514 PLATELET MEMBRANE GLYCOPROTEIN IIB PRECURSOR (GPIIB) (INTEGRIN ALPHA- IIB) (CD41) - HOMO SAPIENS (HUMAN), 1039 aa.
ATPase_associat	cadherin	cadherin	cadherin
CONSER VATIVE	CONSER	CONSER VATIVE	CONSER
Val (652)	Gly (653)	Asp (654)	Asp (655)
Ala	Ala	Glu	Asn
F	<u>5</u>	O .	ප
U	o	ن ن	A
AGCCTTCCGGCAG AAAAAGATGCAG[CT]CCCCCAGACCT TCTCTGTGCTGATT	TACCAGAGGCTGC ATCGGCTGCGCG[C /G]AGAGCAGATGG CGTCGTATTTTGGG	TGGTGGGCGCTCC ACTGTATATGGA[G /C]AGCCGGGCAGA CCGAAAACTGGCC G	CTCTCAACAGGCA GGCACCACCTG[A GJACCTGGATCTG GGCGGAAGCACA G
3906	1138	1238	1893
cg432 <i>77</i> 632	cg40310734	cg40310734	cg40310734
423	424	425	426

9 (9p24)	(11923)	17 (17q25.2)	17 (17925.2)
0	1.80E-203	7.40E-199	7.40E-199
Human Gene SWISSPROT- ID:P98155 VERY LOW- DENSITY LIPOPROTEIN RECEPTOR PRECURSOR (VLDL RECEPTOR) - HOMO SAPIENS (HUMAN), 873 aa.	Human Gene SWISSNEW- ID:P06727 APOLIPOPROTEIN A- IV PRECURSOR (APO-AIV) - HOMO SAPIENS (HUMAN), 396 aa.[pcls:SWISSPROT-ID:P06727 APOLIPOPROTEIN A-IV PRECURSOR (APO-AIV) - HOMO SAPIENS (HUMAN), 396 aa.	Human Gene TREMBLNEW- ID:G2826521 MALTASE- GLUCOAMYLASE (EC 3.2.1.20) - HOMO SAPIENS (HUMAN), 1857 aa.	Human Gene TREMBLNEW- ID:G2826521 MALTASE- GLUCOAMYLASE (EC 3.2.1.20) - HOMO SAPIENS (HUMAN), 1857 aa.
eph	eph	glucoamylase	glucoamylase
CONSER VATIVE	CONSER VATIVE	CONSER VATIVE	CONSER VATIVE
Ala (656)	Lys (657)	Arg (658)	Arg (659)
ĞİŻ	Arg	HIS	His
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<u>ن</u>	ر ن	₹	∢
GGTTACAAGTGTG AATGTAGTCGTG[G /C]CTATCAAATGG ATCTTGCTACTGGC	GCCGAGGACGTGC GTGGCAACCTGA[G /A]GGGCAACACCG AGGGCTGCAGAA G	TACGAGGTGCCCT TGGAGACCCCGC[A /GJTGTCCACAGCC GGGCACCGTCCCC A	GAGGAGCCCTTCG GGGTGATCGTGC[A /G]CCGCCAGCTGG ACGGCCGCGTGCT G
1883	949	1036	1108
cg43982507	cg41554010	cg43299024	cg43299024
427	428	429	430

2		14 (14q11.2)	2 (2q21)
0	6.40E-91	9.90E-70	0
Human Gene SWISSPROT- ID:P98164 LOW-DENSITY LIPOPROTEIN RECEPTOR- RELATED PROTEIN 2 (MEGALIN) (GLYCOPROTEIN 330) - HOMO SAPIENS (HUMAN), 1751 aa (fragment).	Human Gene Similar to SWISSPROT-ID:P38159 HETEROGENEOUS NUCLEAR RIBONUCLEOPROTEIN G (HINRNP G) (GLYCOPROTEIN P43) - HOMO SAPIENS (HUMAN), 437 aa.	Human Gene Similar to SWISSPROT-ID:P16452 ERYTHROCYTE MEMBRANE PROTEIN BAND 4.2 (P4.2) (PALLIDIN) - HOMO SAPIENS (HUMAN), 690 aa.	Human Gene SWISSPROT- ID:P09848 LACTASE- PHLORIZIN HYDROLASE PRECURSOR (EC 3.2.1.108) (EC 3.2.1.62) (LACTASE- GLYCOSYLCERAMIDASE) - HOMO SAPIENS (HUMAN),
glycoprotein	glycoprotein	glycoprotein	hydrolase
CONSER VATIVE	CONSER VATIVE	CONSER VATIVE	CONSER
(660)	Arg (661)	Ile (662)	(663)
116	His	Vai	Ile
O	ט	∢	Đ
\delta	V	ප	V
GACTGACTGGGGA AAGGAACCTAAA[A/CJTCGAGTCTGCC TGGATGAATGGAG A	AGTTATTCTAGAG GATACAGAAATC[A /GJTCGAAGTTCCC GAGAAACTAGGGA G	GGACCAGGGGGCC ATGCTGCTCAAT[G/ AJTCTCAGGCCACG TCAAGGAGAGCGG	TGCTTTTCAGGGCG GAAAACTCTCT[A GJTTGTCCTGCGAG CTGAAGATATCCC
12840	1004	2101	999
cg43285373	cg36834323		cg42359655
431	432	433	434

16 (16q22)	18q21.3
16 (14	
2.00E-220 16	2.20E-149
Leu CONSER hydroxysteroid Human Gene SPTREMBL- (664) VATIVE D:Q13194 14-BETA- HYDROXYSTEROID DEHYDROGENASE TYPE 2 - HOMO SAPIENS (HUMAN), 405 aa.	Human Gene Homologous to SPTREMBL-ID:P91456 SIMILAR TO THE IMMUNOGLOBULIN SUPERFAMILY - CAENORHABDITIS ELEGANS, 1173 aa.
hydroxysteroid	CONSER immunoglob
CONSER	CONSER VATIVE
Leu (664)	Phe (665)
Val	Туг
∢	E-
U	¥
GTGTGGGCCTTGG C TGAACTCTAGCA[C /A]GCGGCTAATGT CTCCTGGTTTGGTC	GGAGATGTGGTCA A TTCCTAGTGATT[A/ TJTTTCAGATAGT GGGAGGAAGCAAC
1331	
435 cg43998672 1331	cg43969028 1133
435	436

	
	0
2.50E-206	
Human Gene SWISSNEW- D:P29466 INTERLEUKIN-1 BETA CONVERTASE PRECURSOR (IL-1BC) (EC 3.4.2.36) (IL-1 BETA CONVERTING ENZYME) (ICE) (INTERLEUKIN-1 BETA CONVERTING ENZYME) (P45) (CASPASE-1) (CASP-1)- HOMO SAPIENS (HUMAN), 404 aa.pcls:SWISSPROT-ID:P29466 INTERLEUKIN-1 BETA CONVERTING ENZYME) (IL-1BC) (EC 3.4.2.3.6) (IL-1 BETA CONVERTING ENZYME) (ICE) (INTERLEUKIN-1 BETA CONVERTING ENZYME) (CASPASE-1) (CASPASE-1) - HOMO SAPIENS (HUMAN), 404 aa.	Human Gene SWISSNEW- ID:P33176 KINESIN HEAVY CHAIN (UBIQUITOUS KINESIN HEAVY CHAIN) (UKHC) - HOMO SAPIENS (HUMAN), 963 aa.lpcls:SWISSPROT-ID:P33176 KINESIN HEAVY CHAIN (UBIQUITOUS KINESIN HEAVY CHAIN) (UKHC) - HOMO SAPIENS (HUMAN), 963 aa.
interfeukin	kinesin
VATIVE	CONSER
Arg (666)	(667)
His	Val
O	4
V	U
AAGGAGAAA AGCTGTTTATCC[A/ GJTTCCATGGGTGA AGGTACAATAAAT	GCCACTGTCTCTTC CAAACCCTTCA[C/ A]GCCTTGTCTTGC TTGTTCTCGTCTA
133	1163
	cg43942537
437	438

	61	19	(11922)
1.80E-113	1.80E-113	1.80E-113	0
Human Gene Homologous to SPTREMBL-ID:Q95368 HLA CLASS I INHIBITORY NK RECEPTOR - HOMO SAPIENS (HUMAN), 455 aa.	Hunan Gene Homologous to SPTREMBL-ID:Q95368 HLA CLASS I INHIBITORY NK RECEPTOR - HOMO SAPIENS (HUMAN), 455 aa.	Human Gene Homologous to SPTREMBL-ID:Q95368 HLA CLASS I INHIBITORY NK RECEPTOR - HOMO SAPIENS (HUMAN), 455 aa.	Human Gene SWISSNEW- ID:P06401 PROGESTERONE RECEPTOR (PR) - HOMO SAPIENS (HUMAN), 933 aa,pcis:SWISSPROT-ID:P06401 PROGESTERONE RECEPTOR (PR) - HOMO SAPIENS (HUMAN), 933 aa.
МНС	МНС	MHC	nucl_recpt
CONSER	CONSER VATIVE	CONSER	CONSER VATIVE
lle (668)	(669)	Asp (670)	(671)
Val	Val	Asn	Val
∢	U	ڻ ت	<u>-</u>
O	<u>්</u>	∀	ව
TTCCAAATGCTGA GCCCAGAGCGTT[G /AJTCTCCTGCCCAT GAGCACCACAGTC	CTGGAACAGTTTC (CTCATTAGCCCT[G/ CJTGACCCCAGCAC ACGCAGGGACCTA	TCATCGCTGGTGCT / CCAAAAAAAA[A GJATGCTGCTGTAA TGAACCAAGAGCC	GGATGCTGTTGCTC C TCCCACAGCCA[G/ TJTGGGCGTTCCAA ATGAAAGCCAAGC
1035	271	823	3434
cg38337333	cg38337333	cg38337333	cg30421838
439	440	441	

	3 (3p21.3)	-	
22	3 (3p2	12	12
4.10E-254	0	1.10E-97	1.10E-97
Human Gene SWISSPROT- ID:Q07869 PEROXISOME PROLIFERATOR ACTIVATED RECEPTOR ALPHA (PPAR- ALPHA) - HOMO SAPIENS (HUMAN), 468 aa.jpcls:SPTREMBL-ID:Q16241 PEROXISOME PROLIFERATOR ACTIVATED RECEPTOR ALPHA - HOMO SAPIENS (HUMAN), 468 aa (fragment).	Human Gene SWISSPROT- ID:P40692 MUTL PROTEIN HOMOLOG \(\) (DNA MISMATCH REPAIR PROTEIN MLH1) - HOMO SAPIENS (HUMAN), 756 aa.	Human Gene Similar to SWISSPROT-ID:P01118 TRANSFORMING PROTEIN P21/K-RAS 2B - HOMO SAPIENS (HUMAN), 188 aa.	Human Gene Similar to SWISSPROT-ID:P01118 TRANSFORMING PROTEIN P21/K-RAS 2B - HOMO SAPIENS (HUMAN), 188 aa.
nucl_recpt	nuclease	oucogene	oncogene
CONSER VATIVE	CONSER VATIVE	CONSER VATIVE	CONSER VATIVE
Val (672)	Val (673)	Glu (674)	Gln (675)
Ala	<u> </u>	Gln	Olu
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	TCTCGACTAACAG CATTTCCAAAGA[T/ CJGGAGCGAATATT GTCCACGGTTGAG	GAGTGCCTTGACG ATACAGCTAATT[C/ G]AGAATCATTTTG TGGACGAATATGA	AAGAAGTTATGGA ATTCCTTTTATT[G/ C]AAACATCAGCAA AGACAAGACAGGG
1019	1860	194	548
	cg43991813	cg42904626	cg42904626
443	444	445	446

3 (3p14.2)	19	6 (6p12)
0	1.40E-79	0
Human Gene SWISSPROT- ID:P23470 PROTEIN-TYROSINE PHOSPHATASE GAMMA PRECURSOR (EC 3.1.3.48) (R- PTP- GAMMA) - HOMO SAPIENS (HUMAN), 1445 aa.	Human Gene Similar to SPTREMBL-ID:Q61469 PHOSPHATIDIC ACID PHOSPHATASE - MUS MUSCULUS (MOUSE), 283 aa.	Human Gene SWISSNEW- ID:P25205 DNA REPLICATION LICENSING FACTOR MCM3 (DNA POL YMERASE ALPHA HOLOENZYME-ASSOCIATED PROTEIN PI) (RLF BETA SUBUNIT) (P102 PROTEIN) - HOMO SAPIENS (HUMAN), 808 aa_pcls:SWISSPROT-ID:P25205 DNA REPLICATION LICENSING FACTOR MCM3 (DNA POL YMERASE ALPHA HOLOENZYME-ASSOCIATED PROTEIN PI) (RLF BETA SUBUNIT) (P102 PROTEIN) - HOMO SAPIENS (HUMAIN), 808 aa.
phosphatase	phosphatase	polymerase
CONSER	CONSER VATIVE	CONSER
Val (676)	Ala (677)	Val (678)
Ala	Val	Leu
E	U	ප
U	l⊶	ပ
GCCGCCTCAGCCA GCAAGCAGGCGG CTJTAGGCCACAGTCC TAGCCACCACAGA G	GGGATGTACTGCA TGGTGTTCTTGG[T/ CJGCTGTATGTGCA GGCACGACTCTGT	TCAGGTGGTGGGA ACCTACCGTTGC[C/ GJTCCTGGAAAGA AGGGAGGCTACAC
2845	282	807
1	cg43272594	cg43958858
447	448	

7 (7q21.3)		12 (12q24.2)	12 (12q24.2)
1.20E-247	0	0	0
Human Gene SPTREMBL- ID:Q15113 PROCOLLAGEN C- PROTEINASE ENHANCER PROTEIN PRECURSOR - HOMO SAPIENS (HUMAN), 449 aa.	Human Gene SWISSPROT- ID:P54296 M-PROTEIN (165 KD ITITN-ASSOCIATED PROTEIN) (165 KD CONNECTIN- ASSOCIATED PROTEIN) HOMO SAPIENS (HUMAN), 1465 aa.	Human Gene SWISSPROT- D:P29475 NITRIC-OXIDE SYNTHASE, BRAIN (EC 1.14.13.39) (NOS, TYPE I) (NEURONAL NOS) (NNOS) - HOMO SAPIENS (HUMAN), 1434 aa.	Human Gene SWISSPROT- ID:P29475 NITRIC-OXIDE SYNTHASE, BRAIN (EC 1.143.39) (NOS, TYPE I) (NEURONAL NOS) (NNOS)- HOMO SAPIENS (HUMAN), 1434 ag.
protease	struct	synthase	synthase
CONSER VATIVE	CONSER VATIVE	CONSER	CONSER
Ser (679)	Asp (680)	(681)	Ala (682)
TL.	Asn	Val	Gly
E	_ව	ပ	ပ
<	∀	U	o ·
GTACAGCGGGCGG GCCACCTCGGGC[A TICTGAGCACCAA TTTTGCGGGGGGGC G	GGATGCTGGAGAG TGGATCACTGTC[A/ GJATCAGACGACA ACAGCCAACCGTT A	GATTCCTCCAGAG CTGGTGTTGGAA[G /CJTTCCCATCAGGC ACCCCAAGTTTGA	AAGTTTGAGTGGT TCAAGGACCTGG[G /C]GCTGAAGTGGT ACGGCCTCCCCGC C
540	2745	2337	2380
			cg40388639
450	451	452	453

91	
7.70E-79	7.70E-79
Human Gene Similar to SWISSNEW-ID:P39062 ACETYL-COENZYME A SYNTHETASE (EC 6.2.1.1) (ACETATE-COA LIGASE) (ACYL-ACTIVATING ENZYME) (ACETYL-COA SYNTHASE) - BACILLUS SUBTILLIS, 572 aa.lpcls:SWISSPROT-ID:P39062 ACETYL-COENZYME A SYNTHETASE (EC 6.2.1.1) (ACETATE-COA LIGASE) (ACYL-ACTIVATING ENZYME) (ACETYL-COA SYNTHASE) - BACILLUS SYNTHASE) - BACILLUS SYNTHASE) - BACILLUS SYNTHASE) - BACILLUS SYNTHASE) - BACILLUS	Human Gene Similar to SWISSNEW-ID:P39062 ACETYL-COENZYME A SYNTHETASE (EC 6.2.1.1) (ACETATE-COA LIGASE) (ACYL-ACTIVATING ENZYME) (ACETYL-COA SYNTHASE) - BACILLUS SUBTILIS, 572 aa.lpcls:SWISSPROT-ID:P39062 ACETYL-COENZYME A SYNTHETASE (EC 6.2.1.1) (ACETATE-COA LIGASE) (ACETATE-COA LIGASE) (ACETATE-COA LIGASE) (ACETATE-COA LIGASE) (ACETATE-COA LIGASE) SYNTHASE) - BACILLUS SYNTHASE) - BACILLUS SYNTHASE) - BACILLUS SYNTHASE) - BACILLUS
synthase	synthase
CONSER VATIVE	CONSER
(683) (683)	(684)
Ala	FE CONTRACTOR OF THE CONTRACTO
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lo lo	V
AATTTCTATATCAC TGGGGACAGAG[C/ GJATATATGGATAA AGATGGTATTTC	TGGAACAAGTGGA TATCCGAAAATGIA AJCTGCACACACCC ACAGCAGTTTTGG
1524	698
cg43124627	cg43124627
454	455

	-	3 (3q21)	∞
7.40E-65	7.00E-172		5.50E-89 8
Human Gene Similar to SWISSNEW-ID:P39062 ACETYL-COENZYME A SYNTHETASE (EC 6.2.1.1) (ACETATE-COA LIGASE) (ACYL-ACTIVATING ENZYME) (ACETYL-COA SYNTHASE) - BACILLUS SUBTILLIS, 572 aa.jpcis:SWISSPROT-ID:P39062 ACETYL-COENZYME A SYNTHETASE (EC 6.2.1.1) (ACETATE-COA LIGASE) (ACYL-ACTIVATING ENZYME) (ACETYL-COA SYNTHASE) - BACILLUS SYNTHASE) - BACILLUS SYNTHASE) - BACILLUS	Human Gene SWISSPROT- ID:P33032 MELANOCORTIN-5 RECEPTOR (MC5-R) (MC-2) - HOMO SAPIENS (HUMAN), 325 aa.	Human Gene Similar to SPTREMBL-ID:Q89609 G PROTEIN-COUPLED RECEPTOR - EQUINE HERPESVIRUS TYPE 2 (EHV-2), 383 aa.	Human Gene Similar to TREMBLNEW-ID:G2653845 TNF RECEPTOR-RELATED RECEPTOR FOR TRAIL - HOMO SAPIENS (HUMAN), 386 aa.
synthase	tm7	m.7	infreceptor
CONSER VATIVE	CONSER		CONSER
lle (685)	Phe (686)	Tyr (687)	Val (688)
Val	T _Y	Phe	Ala
Α	E-	<u> </u>	E-
O	⋖	∢	U
AGGAGGTGGTG AAGGCATTTGTGG AATCCTGCCTCGC AGTTCCTGTCCCA	GTGATGGACCCTC TCATATATGCCT[A/ TJCCGCAGCCAAGA GATGCGGAAGACC	TTCCATCTGAGGTT TATAAACCACG[A/ TJATTCAGGCAAAG TGGCCAGAATGGC	CAAGACCTAGCTC CCCAGCAGAGAG[CT]GGCCCCACAA CAAAAGAGGTCCA GC
·	0601	964	344
			cg4333558
456	457	458	459

12	9 (9434)
1.70E-177	6.50E-192
Human Gene SPTREMBL- ID:Q07279 TRANSCRIPTION FACTOR NF-E2 - MUS MUSCULUS (MOUSE), 373 aa.	Human Gene SWISSPROT- ID:P16442 FUCOSYLGL YCOPROTEIN ALPHA-N- ACETYLGALACTOSAMINYLT RANSFERASE (EC 2.4.1.40) (HISTO-BLOOD GROUP A TRANSFERASE) / FUCOSYLGL YCOPROTEIN 3- ALPHA- GALACTOSYLTRANSFERASE (EC 2.4.1.37) (HISTO-BLOOD GROUP B TRANSFERASE) (B
CONSER transcriptfactor VATIVE	transferase
CONSER	CONSER VATIVE
Glu (689)	(690) (690)
Qlu	Gly
ტ	O
D D	O .
GACAGAGCTGTAC C CGTGACATTTTC[C/ GJAGCACCTTCGGG ATGAATCAGGCAA	GAGGGCGATTTCT ACTACCTGGGGGG /C]GTTCTTCGGGGG GTCGGTGCAAGAG
	008
cg43998970 1347	cg2537639
460	461

9	6 (6p21.3)	6	
·	0	0	o
Human Gene SWISSPROT- ID:Q03518 ANTIGEN PEPTIDE TRANSPORTER 1 (APT1) (PEPTIDE TRANSPORTER TAANSPORTER PSF1) (PEPTIDE SUPPLY FACTOR 1) (PSF-1) (PEPTIDE TRANSPORTER INVOLVED IN ANTIGEN PROCESSING 1)- HOMO SAPIENS (HUMAN), 748 aa.	Human Gene SPTREMBL- ID:Q28437 ABC-TRANSPORTER - GORILLA GORILLA GORILLA (LOWLAND GORILLA), 703 aa.	Human Gene SPTREMBL- ACC:014914 NEURONAL MUNC18-1 BINDING PROTEIN - HOMO SAPIENS (HUMAN), 837 aa.	Human Gene SWISSPROT- ACC:P29374 RETINOBLASTOMA BINDING PROTEIN 1 (RBBP-1) - Homo sapiens (Human), 1257 aa.
transport	transport	UNCLASSIFIE D	UNCLASSIFIE D
CONSER	CONSER	CONSER VATIVE	CONSER VATIVE
(691)]le (692)	Val (693)	Val (694)
110	Val	Ala	Leu
0	4	H	ပ
A	ڻ ن	ပ	Ð
AGTGTCCTCACC ATGGTCACCCTG[A /G]TCACCCTGCCTC TGCTTTTCCTTCT	CCTGGAACGCGCC TTGTACCTGCTC[G/ AJTAAGGAGGTG CTGCACTTGGGGG	GAGCACGAGGAAG CCATGAATGCGG[C /I]CTACTCAGGCTA CGTCTACACGCAC	AGATACTTICTATA AGCAGTTTTTA[G/C JATTGTAGGAAGCA GCTGAATTCAAA
1552	1424	730	3568
			cg44018598
462	463	464	465

	T			
	11	1 (1942)	22 (22q11.2)	6 (6q23)
0	0	3.90E-257	3.30E-228	1.30E-171
Human Gene SWISSPROT- ACC:Q15046 LYSYL-TRNA SYNTHETASE (EC 6.1.1.6) (LYSINETRNA LIGASE) (LYSRS) (KIAA0070) - Homo sapiens (Human), 597 aa.	Human Gene SWISSPROT- ACC:P42694 HYPOTHETICAL PROTEIN KIAA0054 - Homo sapiens (Human), 1942 aa.	Human Gene SWISSPROT- ACC:P01019 ANGIOTENSINOGEN PRECURSOR - Homo sapiens (Human), 485 aa.	Human Gene SWISSPROT- ACC.P20062 TRANSCOBALAMIN II PRECURSOR - Homo sapiens (Human), 427 aa.	Human Gene SWISSPROT- ACC:P05089 ARGINASE 1 (EC 3.5.3.1) (LIVER-TYPE ARGINASE) - Homo sapiens (Human), 322 aa.
UNCLASSIFIE D	UNCLASSIFIE D	UNCLASSIFIE D	UNCLASSIFIE D	UNCLASSIFIE D
CONSER	CONSER VATIVE	CONSER	CONSER VATIVE	CONSER VATIVE
Ser (695)	His (696)	Glu (697)	Leu (698)	Gln (699)
Thr	Arg	Glu	Ile	Glu
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U	ပ	ပ	A	U
ACACTGGAAAGCA CAACAGTTGGCA[C /G]TTCTGTCTAGAA AATAATAATTGCA	AACGCTGCCCTGA CTGAGAAAGGCA[CTJGATGCTCGCTC CACTGCTGGAACC G	CATCCAGGACAAC TTCTCGGTGACT[C/ G]AAGTGCCCTTCA CTGAGAGCGCCTG	CTTCAACCTGGTC GGAGACAACGG[A/ CJTCACCATGGCCA TCAGAACAGTGCG	CTGATTCTTCCGTT CTTCTTGACTT[C/G JTGCCACCTTGCCA GCCAGCTGCTCG
	1622	1381	996	1148
cg44926796	cg43055918	cg43966985		cg43918484
466	467	468	469	470

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		6 (6q25.3)	
9.60E-148	9.60E-148	5.70E-124	7.20E-91
Human Gene Homologous to SWISSNEW-ACC:Q12846 SYNTAXIN 4 - Homo sapiens (Human), 297 aa.	Human Gene Homologous to SWISSNEW-ACC:Q12846 SYNTAXIN 4 - Homo sapiens (Human), 297 aa.	Human Gene Homologous to SWISSNEW-ACC:P04179 SUPEROXIDE DISMUTASE [MN] PRECURSOR (EC 1.15.1.1) - Homo sapiens (Human), 222 aa.	Human Gene Similar to SWISSNEW-ACC:P01185 VASOPRESSIN-NEUROPHYSIN 2-COPEPTIN PRECURSOR [CONTAINS: ARG-VASOPRESSIN; NEUROPHYSIN 2 (NEUROPHYSIN-II); COPEPTIN] - Homo sapiens (Human), 164 aa.
UNCLASSIFIE D	UNCLASSIFIE D	UNCLASSIFIE D	UNCLASSIFIE D
CONSER	CONSER	CONSER VATIVE	CONSER VATIVE
Val (700)	Asp (701)	Gln (702)	Val (703)
Ala	n G G	Glu	Gly
H	<u>-</u>	υ	<u> </u>
ن	Ö	_ල	ڻ ن
ACGGCCCTGGAGA ACCAGAAGAAGG CTJGAGGAAGAAG AAAGTCTTGATTG CC	GGATGGTGTCTGA TGAGGAGTTGGA[G /TJCAGATGCTGGA CAGTGGGCAAAGC G	TTGGGGTTGGCTTG C GTTTCAATAAG[G/ CJAACGGGGACACT TACAAATTGCTGC	GTGACCGAGCCCG AGTGCCGCGAGG[GAJCTTTCACCGCC GCGCCCGCGCAG
1009	725	921	1094
	cg43942977	cg43943361	og25236776
471	472	473	474

		7		
7.20E-91	5.70E-75	1.30E-66	1.60E-54	1.60E-54
Human Gene Similar to SWISSNEW-ACC:P01185 VASOPRESSIN-NEUROPHYSIN 2-COPEPTIN PRECURSOR [CONTAINS: ARG- VASOPRESSIN; NEUROPHYSIN 2 (NEUROPHYSIN-II); COPEPTIN] - Homo sapiens (Human), 164 aa.	Human Gene Similar to REMTREMBL-ACC:G292791 T- CELL RECEPTOR BETA PRECURSOR - HOMO SAPIENS (HUMAN), 145 aa (fragment).	Human Gend Similar to REMTREMBL-ACC:G2104755 T CELL RECEPTOR V-BETA 23 - HOMO SAPIENS (HUMAN), 129 aa (fragment).	Human Gene Similar to REMTREMBL-ACC:G33509 T CELL RECEPTOR - HOMO SAPIENS (HUMAN), 118 aa (fragment).	Human Gene Similar to REMTREMBL-ACC:G33509 T CELL RECEPTOR - HOMO SAPIENS (HUMAN), 118 aa (fragment).
UNCLASSIFIE D	UNCLASSIFIE D	UNCLASSIFIE D	UNCLASSIFIE D	UNCLASSIFIE D
CONSER VATIVE	CONSER VATIVE	CONSER	CONSER VATIVE	CONSER
Val (704)	(705)	His (706)	Tie (707)	Asp (708)
Gly	Vai	Arg	Vai	Glu
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<u>ე</u>	ڻ ن	ပ	Ů	ິ ອ
CAGTGCCTCCCTG CGGCCCGGGG(a/ TJCAAAGGCCGCTG CTTCGGGCCCAGC	GGCCAACTCTGCT ATGGACACCAGA[G/CJTACTCTGCTGT GCGGTCATCTGTCT	GCCTGGAACACCA GGCTCCTCTGCC[G/ AJTGTCATGCTTTG TCTCCTGGGAGCA	AGCCACCCAGACC GGAGACTCGGCC[G /A]TCTACCTCTGTG CTGTGGAGGCCTA	CGGCCGTCTACCTC TGTGCTGTGGA[G/ CJGCCTATTCTAAC GACTACAAGCTCA
	30	253	519	539
og25236776	cg38899722	cg11753818		cg2526759
475	476	477	478	479

	13 (13q14.3)
2.10E-52	0
CONSER UNCLASSIFIE Human Gene Similar to SWISSPROT-ACC:P01286 SOMATOLIBERIN PRECURSOR (GROWTH HORMONE- RELEASING FACTOR) (GRF) (GROWTH HORMONE- RELEASING HORMONE- RELEASING HORMONE) (GHRH) (SOMATOCRININ) - Homo sapiens (Human), 108 aa.	ATPase_associat Human Gene SWISSPROT- ed ID:P35670 COPPER- TRANSPORTING ATPASE 2 (EC 3.6.1.36) (COPPER PUMP 2) (WILSON DISEASE- ASSOCIATED PROTEIN) HOMO SAPIENS (HUMAN), 1465 aa.
UNCLASSIFIE D	ATPase_associat ed
CONSER VATIVE	NON- CONSER VATIVE
Asp (709)	Gly (710)
Glu	Arg
<u>-</u>	<u>ව</u>
O	A
CAGAACAAAGCA G AATGGAATTGGA[G //JAGCATCCTGGTG GCCCTGCTGCAGA	GAAACCGGAAGC A ACTGTAATTGCGĮA IGJGGTCTATAAAT GCACATGGCTCTG T
	3110
cg1902363 368	cg432 <i>77</i> 632
480	481

X (Xq12)	3 (3q21)	17 (17q21.3 2)
0	8.20E-288	0
ATPase_associal Human Gene SWISSNEW- ID:Q04656 COPPER- TRANSPORTING ATPASE 1 (BC 3.6.1.36) (COPPER PUMP 1) (MENKES DISEASE- ASSOCIATED PROTEIN) - HOMO SAPIENS (HUMAN), 1500 aa, [pcls:SWISSPROT- ID:Q04656 COPPER- TRANSPORTING ATPASE 1 (EC 3.6.1.36) (COPPER PUMP 1) (MENKES DISEASE- ASSOCIATED PROTEIN) - HOMO SAPIENS (HUMAN), 1500 aa.	Human Gene SWISSPROT- ID:P05166 PROPIONYL-COA CARBOXYLASE BETA CHAIN PRECURSOR (EC 6.4.1.3) (PCCASE) (PROPANOYL- COA:CARBON DIOXIDE LIGASE) - HOMO SAPIENS (HUMAN), 539 aa.	Human Gene SWISSPROT- ID:P08514 PLATELET MEMBRANE GLYCOPROTEIN IIB PRECURSOR (GPIIB) (INTEGRIN ALPHA- IIB) (CD41) - HOMO SAPIENS (HUMAN), 1039 aa.
ATPase_associat	biotindep	cadherin
NON- CONSER VATIVE	NON- CONSER VATIVE	NON- CONSER VATIVE
(711)	Ser (712)	Ala (713)
臣	Pro	Pro
E-	<u> </u>	_ව
ပ	O	U
TGTATTCCTGTAAT GGGGCTGATGA[C/ T]ATATATGTGT TATGGACCACCAC	GCCCCTGAGCAGT CAGGACCCGGCT[C /TJCCGTCCGTGAGT GCCACGATCCCAG	GGAGTGGGTGCTG CTGCTCTTGGGA[C/ GJCTTGTGCTGCCC CTCCAGCCTGGGC
2306	929	267
og43252813	cg43920913	cg40310734
482	483	484

17 (17q21.3 2)	1 (1423)
0	1.00E-218
Human Gene SWISSPROT- ID:P08S14 PLATELET MEMBRANE GLYCOPROTEIN IIB PRECURSOR (GPIIB) (INTEGRIN ALPHA- IIB) (CD41) - HOMO SAPIENS (HUMAN), 1039 aa.	Human Gene SWISSPROT- ID:P14151 L-SELECTIN PRECURSOR (L-YMPH NODE HOMING RECEPTOR) (LEUKOCYTE ADHESION MOLECULE-1) (LAM-1) (LEUKOCYTE SURFACE ANTIGEN LEU-8) (ITQ1) (GP90- MEL) (LEUKOCYTE- ENDOTHELIAL CELL ADHESION MOLECULE 1) (LECAMI) (CD62L) - HOMO SAPIENS (HUMAN), 372 aa.
cadherin	cadherin
NON- CONSER VATIVE	NON- CONSER VATIVE
Ala (714)	(715)
Pro	Phe
ර	ပ
C)	<u></u>
CGTGTCCCCCTCC C CCTATGCGGTG[C/ G]CCCCGCTCAGCC TGCCCCGAGGGGA	GGGGTACTATGGG CCCCAGTGTCAGT/ CJTTGTGATTCAGT GTGAGCCTTTGGA
	777
cg40310734 3111	cg43956560
485	486

1 (1923)	4	
1.00E-218	7.00E-172	0
Human Gene SWISSPROT- ID:P14151 L-SELECTIN PRECURSOR (LYMPH NODE HOMING RECEPTOR) (LEUKOCYTE ADHESION MOLECULE-1) (LAM-1) (LEUKOCYTE SURFACE ANTIGEN LEU-8) (TQ1) (GP90- MEL) (LEUKOCYTE- ENDOTHELIAL CELL ADHESION MOLECULE 1) (LECAMI) (CD62L) - HOMO SAPIENS (HUMAN), 372 aa.	Human Gene SWISSPROT- ID:P21815 BONE SIALOPROTEIN II PRECURSOR (BSP II) (CELL-BINDING SIALOPROTEIN) (INTEGRIN- BINDING SIALOPROTEIN) - HOMO SAPIENS (HUMAN), 317 aa.	Human Gene SWISSPROT- ID:P21817 RYANODINE RECEPTOR, SKELETAL MUSCLE (SKELETAL MUSCLE CALCIUM RELEASE CHANNEL) - HOMO SAPIENS (HUMAN), 5032 aa.
cadherin	cadherin	calcium_channel
NON- CONSER VATIVE	NON- CONSER VATIVE	NON- CONSER VATIVE
Ser (716)	Gly (717)	Cys (718)
Pro	Arg	Arg
 	<u>ဗ</u>	<u>[</u>
υ	∢	ن. ن
GCTGGGTACCATG GACTGTACTCAC[C/ T]CTTTGGGAAACT TCAGCTTCAGCTC	TGCAGAAGGCACC ACAGAGACCGGA[A/G]GGCAGGGCAA GGGCACCTCGAAG AC	GTGTGTGTGTAAT GGTGTGGCTGTA[C /T]GCTCCAACCAA GATCTTATTACTGA
837	753	1945
		cg43977436
487	488	489

2	9	9_	X (Xq22)
Human Gene SWISSPROT- ID:P38435 VITAMIN K- DEPENDENT GAMMA- CARBOXYLASE (EC 64) (GAMMA-GLUTAMYL CARBOXYLASE) - HOMO SAPIENS (HUMAN), 758 aa.	Human Gene SWISSPROT- 1D:Q03692 COLLAGEN ALPHA 1(X) CHAIN PRECURSOR - HOMO SAPIENS (HUMAN), 680 aa.	Human Gene SWISSPROT- 1D: Q03692 COLLAGEN ALPHA 1(X) CHAIN PRECURSOR - HOMO SAPIENS (HUMAN), 680 aa.	Human Gene SPTREMBL- ID:Q12823 A TYPE IV COLLAGEN - HOMO SAPIENS (HUMAN), 1690 sa (fragment).
carboxylase	collagen	collagen	collagen
NON- CONSER VATIVE	NON- CONSER VATIVE	NON- CONSER VATIVE	NON- CONSER VATIVE
Arg (719)	Gly (720)	Met (721)	Thr (722)
Gin	Пр	걘	lie
b	ပ	<u>-</u>	D_
∢	V	ပ	V .
CGGAAGCTGGTGT // CCTACTGCCCCC[A/ GJAAGGTTGCAACA ACTGTTGCCCCTC	CCAGGGCCTCCAG GTCCAAGAGGCCIA /CJCTCTGGAGAGG CTGGTCTTCCAGG G	GTGTTTTACGCTGA ACGATACCAAA[C/ TJGCCCACAGGCAT AAAAGGCCCACTA	TCCAGGAATACCA GGTCTGCCTGGT[A/ GJTTCCTGGAACAA GAGGATTAAAAGG
1130	1595	9/1	2855
cg43280376	cg42201364	cg42201364	cg40339378
490	491	492	493

1 (1p32)	I (1p32)	5 (5p13)	I (1p21)
0	0	0	5.00E-304
Human Gene SWISSNEW- ID:P07358 COMPLEMENT COMPONENT C8 BETA CHAIN PRECURSOR - HOMO SAPIENS (HUMAN), 591 aa.lpcis:SWISSPROT-ID:P07358 COMPLEMENT COMPONENT C8 BETA CHAIN PRECURSOR - HOMO SAPIENS (HUMAN), 591 aa.	Human Gene SWISSPROT- D:P07357 COMPLEMENT COMPONENT C8 ALPHA CHAIN PRECURSOR - HOMO SAPIENS (HUMAN), 584 aa	Human Gene TREMBLNEW- ID:G386348 COMPLEMENT C6 - HOMO SAPIENS, 941 aa.	Human Gene SWISSPROT- ID:P09603 MACROPHAGE COLONY STIMULATING FACTOR-1 PRECURSOR (CSF-1) (MCSF) - HOMO SAPIENS (HUMAN), 554 aa.
complement	complement	complement	csf.
NON- CONSER VATIVE	NON- CONSER VATIVE	NON- CONSER VATIVE	NON- CONSER VATIVE
(723) (723)	Gln (724)	Ala (725)	Pro (726)
Arg	Lys	Glu	Leu
g	O	ပ	ပ
4	4	∀	[—
AGACTGTGTTACC AACAGACCATGC[A /G]GAAGTCAAGTG CGATGTGAAGGCT T	CTCCAGTTCTACAA CTTGTGTAAGG[A/ C]AAGCACAGTGTG GACAGGATTTCCA	CAGTTTGGGGGGAC A AGCCATGCACTG[A /C]GCCTCTGGTAGC CTTTCAACCATGC	CCAGGCTCTCCCA GGATCTCATCAC[T/ CJGCGCCCCAGGG CCTCAGCAACCCC
909	414	233	1347
cg43063256			cg21644442
494	495	496	497

V	
1.10E-77	70E-52
Human Gene Similar to SWISSNEW-ID:P08700 INTERLEUKIN-3 PRECURSOR (IL-3) (MULTIPOTENTIAL COLONY-STIMULATING FACTOR) (HEMATOPOIETIC GROWTH FACTOR) (MAST-CELL GROWTH FACTOR) (MCGF) - HOMO SAPIENS (HUMAN), 152 aa.pcls:SWISSPROT-ID:P08700 INTERLEUKIN-3 PRECURSOR (IL-3) (MULTIPOTENTIAL COLONY-STIMULATING FACTOR) (HEMATOPOIETIC GROWTH FACTOR) (P-CELL STIMULATING FACTOR) (MAST-CELL GROWTH FACTOR) (MCGF) - HOMO SAPIENS (HUMAN), 152 aa.	Human Gene Similar to SWISSPROT-ID:P21592 CYTOCHROME C OXIDASE ASSEMBLY PROTEIN COX10 PRECURSOR - SACCHAROMYCES CEREVISIAE (BAKER'S YEAST), 462 aa.
ાં	cytochrome
NON- CONSER VATIVE	NON- CONSER VATIVE
(727)	His (728)
Pro	Tyr
[-	O
U	V
CCAAGCTCCCATG ACCCAGACGC //JCCTTGAAGACA AGCTGGGTTAACT G	TCCACGTAGAAGC GGAAGCCGAGGT[A/G]GGAGATGTAC GCATTGATGGGAA GG
279	1651
cg2753430	cg43923204
498	499

22	(1p36.2)	4 (4922)
2.40E-52	8.80E-78	1.30E-209
Human Gene Similar to SPTREMBL-ID:000761 CYTOCHROME OXIDASE SUBUNIT VIA HEART ISOFORM PRECURSOR (EC 1.9.3.1) (CYTOCHROME-C OXIDASE) (CYTOCHROME A(3)) (CYTOCHROME AA(3))- HOMO SAPIENS (HUMAN), 97 aa.	Human Gene Similar to SWISSPROT-ID:P32320 CYTIDINE DEAMINASE (EC 3.5.4.5) (CYTIDINE AMINOHYDROLASE) - HOMO SAPIENS (HUMAN), 146 aa.pcls:TREMBLNEW- ID:E1228801 CYTIDINE DEAMINASE (EC 3.5.4.5) - HOMO SAPIENS (HUMAN), 146 aa.	Human Gene SWISSNEW- ID:P08319 ALCOHOL DEHYDROGENASE CLASS II PI CHAIN (EC 1.1.11)- HOMO SAPIENS (HUMAN), 391 aa.pcls:SWISSPROT-ID:P08319 ALCOHOL DEHYDROGENASE ALCOHOL DEHYDROGENASE ALCASS II PI CHAIN (EC 1.1.1.1)- HOMO SAPIENS (HUMAN), 391 aa.
cytochrome	deaminase	dehydrogenase
NON- CONSER VATIVE	NON- CONSER VATIVE	NON- CONSER VATIVE
Gln (729)	Gln (730)	Gly (731)
Pro	Lys	End
—	0	ອ
O	4	[
TTGGTAGGGACGG AACTCGGGGCGC[G /T]GGCGGTGGCCC GAGTGGAGATAGG A	GCTGGTTTGCTCC AGGAGGCCAAG[A/ CJAGTCAGCCTACT GCCCTACAGTCA	AAGCATCCGAACA ATCCTCATCTTT[7/ GJGAAGATGCCAG GAGCAATTCGGAA T
174	279	1618
cg44017721	cg41626024	cg43057018
200		502

	1	1 (1p36.13)
3.90E-86	5.50E-57	1.30E-60
Human Gene Similar to TREMBL.NEW-ID:G913312 DNA BINDING PROTEIN MEF2 {CLONE XMEF2A1} - XENOPUS LAEVIS, 516 aa.	Human Gene Similar to SPTREMBL-ID:Q61491 DNA- BINDING PROTEIN - MUS MUSCULUS (MOUSE), 546 aa.	Human Gene Similar to SWISSNEW-ID:Q02535 DNA- BINDING PROTEIN INHIBITOR ID-3 (ID-LIKE PROTEIN INHIBITOR HLH 1R21) (HELIX- LOOP-HELIX PROTEIN HEIR-1) - HOMO SAPIENS (HUMAN), II 9 aa. pcls:SWISSPROT- ID:Q02535 DNA-BINDING PROTEIN INHIBITOR HLH 1R21) (HELIX-LOOP- HELIX PROTEIN HEIR-1) HOMO SAPIENS (HUMAN), 119 aa.
dna_ma_bind	dna ma bind	dna_ma_bind_in hib
NON- CONSER VATIVE	NON- CONSER VATIVE	NON- CONSER VATIVE
Ala (732)	Lys (733)	Thr (734)
Th.	Asn	Ala
O	V V	[
Y	ပ	O
CCGCACCAACGCC L GACATCATCGAG[A /G]CCCTGAGGAAG AAGGGCTTCAAGG G	TCCACGACCGGGT AGAGAACTACAA[CA]CCGCGGCAGC GCAAGCTCCGCAA CC	TCGTTGGAGATGA CAAGTTCCGGAG[C /I]GAGCTCGGCTGT CTGGATGGGAAGG
	2205	707
cg42837709 464	cg43327954	cg43971258
503	504	505

11 (11q23)	
1.805-203	1.90E-178
Human Gene SWISSNEW- ID:P06727 APOLIPOPROTEIN A- IV PRECURSOR (APO-AIV) - HOMO SAPIENS (HUMAN), 396 aa.lpcls:SWISSPROT-ID:P06727 APOLIPOPROTEIN A-IV PRECURSOR (APO-AIV) - HOMO SAPIENS (HUMAN), 396 aa.	Human Gene SWISSNEW- D:Q15166 SERUM PARAOXONASEARYLESTERA SE 3 (EC 3.1.1.2) (EC 3.1.8.1) (PON 3) (SERUM ARYLDHOSPHATASE 3) (A-ESTERASE 3) (AROMATIC ESTERASE 3) - HOMO SAPIENS (HUMAN), 341 aa (fragment) ipcls:SWISSPROT- D:Q15166 SERUM D:Q15166 SERUM PARAOXONASE/ARYLESTERA SE 3 (EC 3.1.1.2) (EC 3.1.8.1) (PON 3) (SERUM ARYLDHAKYLPHOSPHATASE 3) (A-ESTERASE 3) (AROMATIC ESTERASE 3) HOMO SAPIENS (HUMAN), 341 aa (fragment).
ф	esterase
NON- CONSER VATIVE	NON- CONSER VATIVE
His (735)	(736)
Oln Ol	Gly
[-	E-1
<u></u> 5	U
AGCTGGAGCAACA GCAGGAACAGCA[GT]CAGGAGCAGC AGCAGGAGCAGGT GC	GTTTGGCATACCTG GATATTTAAT[CT] JCAGTGGAGATAA AAGACAGCCCACT
1253	1063
cg41554010	cg439 <i>577</i> 43
906	507

	15 (15q15)
1.90E-178	
1.90	9.30E-106
Human Gene SWISSNEW- ID:Q15166 SERUM PARAOXONASEARYLESTERA SE 3 (EC 3.1.1.2) (EC 3.1.8.1) (PON 3) (SERUM ARYLDIAKYLPHOSPHATASE 3) (A-ESTERASE 3) (AROMATIC ESTERASE 3)- HOMO SAPIENS (HUMAN), 341 aa (fragment).pcls:SWISSPROT- ID:Q15166 SERUM PARAOXONASEARYLESTERA SE 3 (EC 3.1.1.2) (EC 3.1.8.1) (PON 3) (SERUM ARYLDIAKYLPHOSPHATASE 3) (A-ESTERASE 3) - HOMO SAPIENS (HUMAN), 341 aa (fragment).	Human Gene Homologous to SWISSPROT-ID:P21781 KERATINOCYTE GROWTH FACTOR PRECURSOR (KGF) (FIBROBLAST GROWTH FACTOR- 7) (FGF-7) (HBGF-7) - HOMO SAPIENS (HUMAN), 194 aa.
esterase	fgf
NON- CONSER VATIVE	NON- CONSER VATIVE
(737)	Glu (738)
Ser	Lys
O	U
V .	V
TATTTAATCCAGT GGAGGTAAAAG[A/ CJCAGCCACTAGG AAGTATATCAATA	AAGTGAATTCTAT CTTGCAATGAAC[A /G]AGGAAGGAAA CTCTATGCAAAGA A
1079	812
	cg43248101
208	509

	2
<u>s</u>	-
7.40E-80	۵
ETA-	Human Gene \$WISSNEW- D:P40967 MELANOCYTE PROTEIN PMEL 17 PRECURSOR (MELANOCYTE LINEAGE-SPECIFIC ANTIGEN GP100) (MELANOMA- ASSOCIATED ME20 ANTIGEN) (ME20)M/ME20-S) (95 KD M/ME20-S) (95 KD MELANOCYTE-SPECIFIC SECRETED GLYCOPROTEIN)- HOMO SAPIENS (HUMAN), 661 aa.jecis:SWISSPROT-ID:P40967 aa.jecis:SWISSPROT-ID:P40967 MELANOCYTE PROTEIN PMEL 17 PRECURSOR (MELANOCYTE LINEAGE- SPECIFIC ANTIGEN GP100) (MELANOMA-ASSOCIATED ME20 ANTIGEN) (ME20M) (MELANOMA-ASSOCIATED ME20 ANTIGEN) (ME20M) (MELANOMA-SSOCIATED ME20 ANTIGEN) (ME20M) (MECOMO SAPIENS (HUMAN), 661 aa.
glucuronidase	glycoprotein
NON- CONSER VATIVE	NON- CONSER VATIVE
1le (739)	Arg (740)
Arg	Pro
<	ပ
U	ර
GATGAGCTCTCCA ACCACGTATTTT[C/ AJTGCGTTTTTTGAT CCAGACCCAGATG	CACCAGCAGGTG CCCACGATCAGC[G ACJGAACCTGCCCA AGGCCTGCTTCTTG
332	387
cg43969014	cg43286488
510	511

Human Gene SWISSPROT- ID:Q12889 OVIDUCT-SPECIFIC GLYCOPROTEIN PRECURSOR (OVIDUCTAL GLYCOPROTEIN) (OVIDUCTIN) (ESTROGEN- DEPENDENT OVIDUCT PROTEIN) - HOMO SAPIENS (HUMAN), 678 aa.	Human Gene \$WISSPROT- D:Q12889 OVIDUCT-SPECIFIC GLYCOPROTEIN PRECURSOR (OVIDUCTEN) GLYCOPROTEIN) GUYCOPROTEIN) GOUDUCTIN) (BSTROGEN- DEPENDENT OVIDUCT PROTEIN) - HOMO SAPIENS (HUMAN), 678 aa.	Human Gene SWISSPROT- ID:Q12889 OVIDUCT-SPECIFIC GLYCOPROTEIN PRECURSOR (OVIDUCTAL GLYCOPROTEIN) (OVIDUCTIN) (ESTROGEN- DEPENDENT OVIDUCT PROTEIN) - HOMO SAPIENS (HUMAN), 678 aa.
glycoprotein	glycoprotein	glycoprotein
NON- CONSER VATIVE	NON- CONSER VATIVE	NON- CONSER VATIVE
His (741)	Pro (742)	Thr (743)
Tyr	Set	Met
ى ت	<u>ර</u>	_ව
∢	₹	∢
TTTTCCCCAGGGGT A CACAGACTGAT[A/ G]ACCCACAGAGGT CAGGGTCTTCTGT	GGGGTCACAGACT , GATAACCCACAGIA GGGTCAGGGTCT TCTGTCCAGTGGTC	CTGATGACCCACA GAAGTCATGGTC[A /GJTTGCCCCAGTG ATCTCAGTCTTCTC
·	672	773
cg44004239 663	cg44004239	cg44004239
512	513	514

(11pter)	1 (1921)
1.80E-195	2.00E-183
Human Gene SWISSPROT- ID:P16070 CD44 ANTIGEN PRECURSOR (PHAGOCYTIC GLYCOPROTEIN J) (PGP-1) (HUTCH-1) (EXTRACELLULAR MATRIX RECEPTOR-III) (ECMR-III) (GP90 LYMPHOCYTE HOMING/ADHESION RECEPTOR) (HERMES ANTIGEN) (HYALURONATE RECEPTOR) (HERMES ANTIGEN) (HYALURONATE RECEPTOR) (HERMES ANTIGEN) (HYALURONATE RECEPTOR) (HERMES ANTIGEN) (HYALURONATE RECEPTOR) (HERMES ANTIGEN) (HYALURONATE RECEPTOR) (HERMES ANTIGEN) (HYALURONATE RECEPTOR) (HERMES ANTIGEN) (HYALURONATE RECEPTOR) (HERMES ANTIGEN) (HYALURONATE RECEPTOR) (HERMES ANTIGEN) (TOW44) - HOMO SAPIENS (HUMAN), 742 aa.	Human Gene SWISSNEW- D:P06126 T-CELL SURFACE GLYCOPROTEIN CDIA PRECURSOR (CDIA ANTIGEN) T6/LEU-6) (HTAI THYMOCYTE ANTIGEN) - HOMO SAPIENS (HUMAN), 327 aa. pcls:SWISSPROT-ID:P06126 T-CELL SURFACE GLYCOPROTEIN CDIA PRECURSOR (CDIA ANTIGEN) (T-CELL SURFACE ANTIGEN) (T-CELL SURFACE ANTIGEN) (T-CELL SURFACE ANTIGEN) (T-CELL SURFACE ANTIGEN) (T-CELL SURFACE ANTIGEN) (T-CELL SURFACE ANTIGEN) (T-CELL SURFACE ANTIGEN) (HUMAN), 327 aa.
glycoprotein	glycoprotein
NON- CONSER VATIVE	NON- CONSER VATIVE
Tyr (744)	(745)
Ser	11 -
<u>H</u>	
<u>5</u>	υ
ATATGTGTCATACT GGGAGGTGTTG[G/ TJATGTGAGGATGT ACACCCCTGTGTT	AAGGAGCCTCTCT CCTTCCATGTCA[C/ TJCTGGATCGCATC CTTTTACAACCAT
1504	
cg43932434	cg40915005
515	516

6.40E-91	6.40E-91
Human Gene Similar to SWISSPROT-ID:P38159 HETEROGENEOUS NUCLEAR RIBONUCLEOPROTEIN G (HNRNP G) (GL YCOPROTEIN P43) - HOMO SAPIENS (HUMAN), 437 aa.	Human Gene Similar to SWISSPROT-ID:P38159 HETEROGENEOUS NUCLEAR RIBONUCLEOPROTEIN G (HINRNP G) (GLYCOPROTEIN P43) - HOMO SAPIENS (HUMAN), 437 aa.
glycoprotein	glycoprotein
NON- CONSER VATIVE	NON- CONSER VATIVE
Cys (747)	Thr (748)
Tyr	Lys
U	O .
 4	<
GTGCTCCCTGATCC TCGTGAAGCAT[A GJTGGTAGCTCAAG TTATGTGGCATCT	AATGCTGCGAAAG ATATGAATGGAA[A /CjGTCTTTGCATGG AAAAGCAATAAAA
1529	329
	cg36834323
518	519
	cg36834323 1529 GTGCTCCCTGATCC A G Tyr Cys NON- glycoprotein Human Gene Similar to SWISSPROT-ID:P38159 TCGTGAAGCAT[A/ GJTGGTAGCTCAAG TTATGTGGCATCT TTATGTGGCATCT (747) CONSER RIBONUCLEOPROTEIN G (HNRNP G) (GLYCOPROTEIN G (HUMAN), 437 aa.

	-		2
6.40E-91	2.50E-80	9.40E-58	2.60E-188
Human Gene Similar to SWISSPROT-ID:P38159 HETEROGENEOUS NUCLEAR RIBONUCLEOPROTEIN G (HNRNP G) (GLYCOPROTEIN P43) - HOMO SAPIENS (HUMAN), 437 aa.	Human Gene Similar to SWISSPROT-ID:P04216 THY-1 MEMBRANE GLYCOPROTEIN PRECURSOR (THY-1 ANTIGEN) (CDW90) (CD90 ANTIGEN) - HOMO SAPIENS (HUMAN), 161 aa.	Human Gene Similar to SWISSPROT-ID:Q05910 CELL SURFACE ANTIGEN MS2 PRECURSOR (EC 3.4.24) (MACROPHAGE CYSTEINE- RICH GLYCOPROTEIN) (CD156 ANTIGEN) - MUS MUSCULUS (MOUSE), 826 aa.	Human Gene SWISSPROT- ID:P28356 HOMEOBOX PROTEIN HOX-D9 (HOX-4C) (HOX-5.2) - HOMO SAPIENS (HUMAN), 342 aa.
glycoprotein	glycoprotein	glycoprotein	homeobox
NON- CONSER VATIVE	NON- CONSER VATIVE	NON- CONSER VATIVE	NON- CONSER VATIVE
Arg (749)	His (750)	Gly (751)	Asp (752)
Ser	Asn	Arg	Ala
O	<u>5</u>	<u>ප</u>	E
V	<u> </u>	V .	O
AAGTCTGAGATCT GCAAGAGGAAGC[A/C]GTGGAGGAAC AAGAGGGTGGCTT CC	GCGGATAAGTAGA GGACCTTCATGT[T/ GJGTATTTGCTGGT GAAGTTGGTTCGG	CTTAGACATACAA TATACTTACCTT[A/ GJGAGGTCACGTAT GTTTGTCCGCACA	GGAGCGAGCGTGG ATCCAGTTCGCG[G /I]CGGGGTTGTTTG GGTCAAGTTGCTG
463	1691	1665	980
		cg42336656	cg42730678
520	521	522	523

	2 (2q21)	
1.10E-123		0
Human Gene Homologous to SWISSPROT-ID:P17509 HOMEOBOX PROTEIN HOX-B6 (HOX-2B) (HOX-2.2) (HU-2) - HOMO SAPIENS (HUMAN), 224 aa.	Human Gene SWISSPROT- D:P09848 LACTASE- PHLORIZIN HYDROLASE PRECURSOR (EC 3.2.1.108) (EC 3.2.1.62) (LACTASE- GLYCOSYLCERAMIDASE)- HOMO SAPIENS (HUMAN),	Human Gene SWISSPROT- ID:Q16666 GAMMA- INTERFERON-INDUCIBLE PROTEIN IFI-16 (INTERFERON- INDUCIBLE MYELOID DIFFERENTIATION TRANSCRIPTIONAL ACTIVATOR) - HOMO SAPIENS (HUMAN), 729 aa.pcis:SFTREMBL-ID:Q16666 IFI16=INTERFERON- INDUCIBLE MYELOID DIFFERENTIATION TRANSCRIPTIONAL ACTIVATOR - HOMO SAPIENS (HUMAN), 729 aa (fragment).
hотеорох	hydrolasc	interferon
NON- CONSER VATIVE	NON- CONSER VATIVE	His NON- (755) CONSER VATIVE
Ser (753)	Ala (754)	His (755) .
]]	Thr	Asp
<u> </u>	ග	Ð
T	V	, U
GCCCTGTGCCTGA CGGAGAGGCAGA[T/G]CAAGATATGG TTCCAGAACCGAC GC	CTGGGCACCATAT A AGGATAGCCCAC[A /G]CCGTCATCAAA GCCCATGCCAGAG T	GTGGAGGGTGCAG GTGAAGTAGCAT[C /G]CACTTCCTTCT CCTCTTTCTTGAT
769	3297	2172
	cg42359655	cg43925670
524	525	526

	21 (21q22.1)	14 (14q32.3)	11 (20p13)	(20p13)
10				
	0	1.40E-262	2.00E-215	2.00E-215
Human Gene SWISSPROT- ID:Q04759 PROTEIN KINASE C, THETA TYPE (EC 2.7.1) (NPKC-THETA) - HOMO SAPIENS (HUMAN), 706 aa.	Human Gene SWISSPROT- ID:Q13627 SERNE/THREONINE-SPECIFIC PROTEIN KINASE MINIBRAIN HOMOLOG (EC 2.7.1) (HP86) (DYRK) - HOMO SAPIENS (HUMAN), 763 aa.	Human Gene SWISSPROT- ID:P31749 RAC-ALPHA SERNE/THREONINE KINASE (EC 27.1) (RAC-PK-ALPHA) (PROTEIN KINASE B) (PKB) (C-AKT) - HOMO SAPIENS (HUMAN), 480 aa.	Human Gene SWISSPROT- ID:P19138 CASEIN KINASE II, ALPHA CHAIN (CK II) (EC 2.7.1.37) - HOMO SAPIENS (HUMAN), AND BOS TAURUS (BOVINE), 391 aa.	Human Gene SWISSPROT- ID:P19138 CASEIN KINASE II, ALPHA CHAIN (CK II) (EC 2.7.1.37) - HOMO SAPIENS (HUMAN), AND BOS TAURUS (BOVINE), 391 aa.
kinase	kinase	kinase	kinase	kinase
NON- CONSER VATIVE	NON- CONSER VATIVE	NON- CONSER VATIVE	NON- CONSER VATIVE	NON- CONSER VATIVE
Leu (756)	Asn (757)	Ser (758)	Val (759)	Arg (760)
Pro	Lys	Gly	Met	Ser
Ę-	H	E	O	U
ပ	_ව	O	E	<u>-</u>
TGCTCCATCAAAA ATGAAGCAAGGCI CTJGCCATGTTTAC CGACACCGGGAAA A	CAAAAGCAAGAAA GTTCTTTGAGAA[G/ TJTTGCCAGATGGC ACTTGGAACTTAA	AGTCCACCGCCGC CTCAGGCCGTGC[C /TjGCTGGCCGAGT AGGAGAACTGGGG G	GGCACTGAAGAAA TCCCTGACATCA[T/ CJATTGGCGCTGCT GACGGGCGTACTG	GGGCTGACAAGGT GCTGATTTTCAC[7/ GJGTGGACAAAGC GTTCCCATCGCTTT
1083	2663	1226	1429	1621
		cg43932396	og43917871	c <u>g</u> 43917871
527	228	529	530	531

(20p13)	11 (20p13)	19 (19q13.1)
2.00E-215	2.00E-215	0
Human Gene SWISSPROT- ID:P19138 CASEIN KINASE II, ALPHA CHAIN (CK II) (EC 2.7.1.37) - HOMO SAPIENS (HUMAN), AND BOS TAURUS (BOVINE), 391 aa.	Human Gene SWISSPROT- ID:P19138 CASEIN KINASE II, ALPHA CHAIN (CK II) (EC 2.7.1.37) - HOMO SAPIENS (HUMAN), AND BOS TAURUS (BOVINE), 391 aa.	Human Gene SWISSNEW- ID:P30530 TYROSINE-PROTEIN KINASE RECEPTOR UFO PRECURSOR (EC 2.7.1.112) (AXL ONCOGENE) - HOMO SAPIENS (HUMAN), 887 aa.jpcls:SWISSPROT-ID:P30530 TYROSINE-PROTEIN KINASE TYROSINE-PROTEIN KINASE RECEPTOR UFO PRECURSOR (EC 2.7.1.112) (AXL ONCOGENE) - HOMO SAPIENS (HUMAN), 887 aa.
kinase	kinase	kinasereceptor
NON- CONSER VATIVE	NON- CONSER VATIVE	NON- CONSER VATIVE
Gly (761)	Phe (762)	Pro (763)
Asp	Leu	Thr
o .	0	ပ
£	[—	<
TTCAATGTTGTATT 1 TGTCAATATAG[T/C]CATATAAATCTTC TGTCCCCAGAAC	TGTAAAATCGAAT ATCATAGTCTGT[T/ G]AACGTCTGGTAC AATTGCTTGAAGT	TCGGCTAGGCAGC CTCCATCCTCAC[A/ C]CCCCTTATCACA TCCGCGTGGCATG
	2096	1107
c <u>g</u> 43917871 1713		cg43322545
532	533	534

19	(19q13.1	_								_			
kinasereceptor Human Gene SWISSNEW-	ID:P30530 TYROSINE-PROTEIN	KINASE RECEPTOR UFO	PRECURSOR (EC 2.7.1.112)	(AXL ONCOGENE) - HOMO	SAPIENS (HUMAN), 887	aa. pcls:SWISSPROT-ID:P30530	TYROSINE-PROTEIN KINASE	RECEPTOR UFO PRECURSOR	(EC 2.7.1.112) (AXL	ONCOGENE) - HOMO SAPIENS	(HUMAN), 887 aa.		
kinasereceptor													
-NON-	_	VATIVE											
Gly	(764)												
Asp		_											
9												 	
Y							_						
1	CCGGCTCGGGG[A/	GICCAGCCAGTGTA	CCTGCCCACTCAG									 	
2116													
535 cg43322545 2116)												
535													

14 (14q21)	6 (6p21.3)
3.90E-139	9.10E-147
Human Gene Homologous to SWISSNEW-ID:P17931 GALECTIN-3 (GALACTOSE-SPECIFIC LECTIN 3) (MAC-2 ANTIGEN) (IGE-BINDING PROTEIN) (35 KD LECTIN) (CARBOHYDRATE BINDING PROTEIN) (GALBY) - HOMO SAPIENS (HUMAN), 249 SAPIENS (HUMAN), 249 SAPIENS (HUMAN), 249 SAPIENS (HUMAN), 249 ANTIGEN) (IGE-BINDING PROTEIN) (GALBOHYDRATE BINDING PROTEIN) (GALBY) (MAC-2 ANTIGEN) (IGE-BINDING PROTEIN) (GALACTOSE-SPECIFIC LECTIN 3) (MAC-2 ANTIGEN) (IGE-BINDING PROTEIN) (GALACTOSIDE-BINDING PROTEIN) (GALBY) - HOMO SAPIENS (HUMAN), 249 aa.	Human Gene Homologous to SWISSPROT-ID:P28068 CLASS II HISTOCOMPATIBILITY ANTIGEN, M BETA CHAIN PRECURSOR - HOMO SAPIENS (HUMAN), 263 aa.
laminin in	МНС
NON- CONSER VATIVE	NON- CONSER VATIVE
His (765)	Glu (766)
Pro .	Ala
E	₹
O .	U
TCCGGGATAAGCT CCAGGTGCTCCA[G //jGGTAGGCGCCT GGAGGTGCCTGTC C	AAGCTTGTCATGC CTCACAGCAGTG[C /A]GCACAAGACTG CCCAGCCCAATGG A
	718
	cg43966144 .
	537

6 (6p21.3)	6 (6p21.3)	19	61	-
9.10E-147	3.70E-134	1.80E-113	1.80E-113	2.30E-71
Human Gene Homologous to SWISSPROT-ID:P28068 CLASS II HISTOCOMPATIBILITY ANTIGEN, M BETA CHAIN PRECURSOR - HOMO SAPIENS (HUMAN), 263 aa.	Human Gene Homologous to SWISSPROT-ID:P06340 HLA CLASS II HISTOCOMPATIBILITY ANTIGEN, DZ ALPHA CHAIN PRECURSOR (MHC DN-ALPHA) - HOMO SAPIENS (HUMAN), 250 aa.	Human Gene Homologous to SPTREMBL-ID:Q95368 HLA CLASS I INHIBITORY NK RECEPTOR - HOMO SAPIENS (HUMAN), 455 aa.	Human Gene Homologous to SPTREMBL-ID:Q95368 HLA CLASS I INHIBITORY NK RECEPTOR - HOMO SAPIENS (HUMAN), 455 aa.	Human Gene Similar to SPTREMBL-ID:P91197 SIMILAR TO LIGAND-GATED IONIC CHANNEL PROTEIN - CAENORHABDITIS ELEGANS, 461 aa.
MHC	МНС	МНС	МНС	misc_channel
NON- CONSER VATIVE	NON- CONSER VATIVE	NON- CONSER VATIVE	NON- CONSER VATIVE	NON- CONSER VATIVE
Thr (767)	(768)	Ala (769)	Ala (770)	Ala (771)
Ile	Ē.	Pro	星	Thr
ပ	[ප	O .	_ව
<u></u>	U	U	4	4
ACTTACACCTGTGT GGTAGAGCACA[T/ CJTGGGGCTCCTGA GCCCATCCTTCGG	GGCCTGGTGGGCT TCCTCGTGGGCA[C/ TJCGTCCTCATCAT CATGGGCACATAT	CTGAGCCCAGAGC GTTGTCTCCTGC[C/ G]CATGAGCACCAC AGTCAGGCCTTGA	AGCCCGGCCGGGC CCCACGGTTCGC[A /G]CAGGAGAGAC GTGACCTTGTCCTG	CCGGCTGTGCTCA GGGGTGTGGGGT[A /G]CGGATACAGAG GAGCGGCTGGTGG A
823	907	1044	424	340
cg43966144	cg42686658	cg38337333	cg38337333	cg42481172
538	539	540	541	542

		7		
8 (8p11.2)	8 (8p11.2)	17 (17q11.2)	X (Xq11)	
8 ®	∞ ∞	55	×	
02-	6			157
6.10E-70	6.10E-70		0	1.40E-157
Human Gene Similar to SPTREMBL-ID:P91197 SIMILAR TO LIGAND-GATED IONIC CHANNEL PROTEIN - CAENORHABDITIS ELEGANS, 461 aa.	AILAR C ANS,	Human Gene SWISSPROT- ID:P20393 V-ERBA RELATED PROTEIN EAR-1 - HOMO SAPIENS (HUMAN), 614 aa.	Human Gene SWISSNEW- ID:P10275 ANDROGEN RECEPTOR - HOMO SAPIENS (HUMAN), 919 aa.lpcis:SWISSPROT-ID:P10275 ANDROGEN RECEPTOR - HOMO SAPIENS (HUMAN), 919 aa.	Human Gene TREMBLNEW- ID:G2935442 RIBONUCLEASE HI - HOMO SAPIENS (HUMAN), 286 aa. pcis: TREMBLNEW- ID:G2935444 RIBONUCLEASE HI - HOMO SAPIENS (HUMAN), 286 aa.
misc channel	misc_channel	nucl_recpt	nucl_recpt	nuclease
NON- CONSER VATIVE	NON- CONSER VATIVE	NON- CONSER VATIVE	NON- CONSER VATIVE	NON- CONSER VATIVE
Leu (772)	Gln (773)	Leu (774)	His (775)	Phe (776)
Ē	Lys	His .	Gln	Leu
<u>. </u>	O	F	O	E .
O	V	∢	<i>.</i> ප	O
GAAGATGCCCTCC TCAGACATGAGTIG TIGAAAGGTTATC AGAAATGGGTCCG Ĉ	AGATGCCCTCCTC AGACATGAGTGG[A/C]AAGGTTATCA GAAATGGGTCCGC CC	GCCTCGGGCTTCC ACTACGGTGTGC[A /I]CGCCTGCGAGG GCTGCAAGGGCTT T	AGCGGGACGGTCC GGAGCAAGCCCA[GCJAGGCAGAGGA GGCGACAGAGGA AA	GTGCCGGGAGTGA GCGATGAGCTGG[C AJITCTGTTCCTGG CCCACAGAGTCGC
238	240	1067	89	16
cg3000465	c <u>g</u> 3000465	cg43249083		cg43323772
543	544	545	546	547

			24.1
	21	12	14 (14q24.1)
-150		16-	101-
6.00E-150	1.10E-97	1.10E-97	8.90E-10
Human Gene Homologous to SPTREMBL-ID:Q13692 BCR/ABL FUSION PROTEIN - HOMO SAPIENS (HUMAN), 284 aa (fragment).	Human Gene Similar to SWISSPROT-ID:P01118 TRANSFORMING PROTEIN P21/K-RAS 2B - HOMO SAPIENS (HUMAN), 188 aa.	Human Gene Similar to SWISSPROT-ID:P01118 TRANSFORMING PROTEIN P21/K-RAS 2B - HOMO SAPIENS (HUMAN), 188 aa.	Human Gene Homologous to SWISSPROT-ID:P18283 GLUTATHIONE PEROXIDASE- GASTROINTESTINAL (EC 1.1.1.9) (GSHPX-GI) (GLUTATHIONE PEROXIDASE- RELATED PROTEIN 2) (GPRP) - HOMO SAPIENS (HUMAN), 190 aa.
oucogene.	oncogene	oncogene	peroxidase
NON- CONSER VATIVE	NON- CONSER VATIVE	NON- CONSER VATIVE	NON- CONSER VATIVE
Phe (777)	Cys (778)	His (779)	(780)
Cys	Gly	ul U	Cys
[H	U	L
o o	b	4	
GGCTATAATCACA ATGGGGAATGGT[G /TJTGAAGCCCAAA CCAAAAATGGCCA A	ATATAAACTTGTG GTAGTTGGAGCT[G /T]GTGGCGTAGGC AAGAGTGCCTTGA C	TGGATATTCTCGAC. ACAGCAGGTCA[A/ CJGAGGAGTACAGT GCAATGAGGGACC	CTGTTCAGGATCTC CTCATTCTGAC[AT]GTTCTCCTGATGT CCAAATTGGTTG
809	155	304	,
cg42732993	cg42904626	cg42904626	cg42691989
548	349	550	551

		(11922)	
7			
		3.20E-302	2.40E-155
Human Gene TREMBINEW- ID:G2262075 IAR/RECEPTOR- LIKE PROTEIN-TYROSINE PHOSPHATASE PRECURSOR- HOMO SAPIENS (HUMAN), 1015 aa.	OSINE (R-	KD N SUNIT BETA)	Human Gene SWISSPROT- ID:P00491 PURINE NUCLEOSIDE PHOSPHORYLASE (EC 2.4.2.1) (INOSINE PHOSPHORYLASE) (PNP) - HOMO SAPIENS (HUMAN), 289 aa.
phosphatase	phosphatase	phosphatase	phosphorylase I
NON- CONSER VATIVE	NON- CONSER VATIVE	NON- CONSER VATIVE	NON- CONSER VATIVE
Pro (781)	Glu (782)	Thr (783)	Ser (784)
Ser.	Gly	e =	Gly
U	H	.	E-
V	U	<i>;</i>	U
AGGTCCTCGCGGA GCTGGGTCCGGG[A /G]CCCGGGAGGGT AGGTCAGCGCAGA C	CTGGGGCACTACT CGGACCTGCTCC[C/ T]CCTGGCGGGCCT GGGGCTGATTGAG	GCACAAGGAACGG AATTGCTGTTGAV GJTTTCTGCTTTAA CAGCATTTGATGC	CTTCGGGGAAAGT TGGGGATTTCAC[C/ TJGTAGTCAAAGAT CTGGGCCTGAGTT
4096	368	3187	1330
cg43917453	cg43947363		cg43996195
552	253	554	555

19 (19413.3)	(2)
(1943)	6 (6p12)
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	·
0	0
Human Gene SWISSNEW- ID:P28340 DNA POLYMERASE DELTA CATALYTIC CHAIN (EC 2.7.7.7) - HOMO SAPIENS (HUMAN), 1107 Balpcis:SWISSPROT-ID:P28340 BAR POLYMERASE DELTA CATALYTIC CHAIN (EC 2.7.7.7) - HOMO SAPIENS (HUMAN), 1107 aa.	N 40 .888 808 40 .88
ARA AIN 1283 AN)	CM3 2520, N, ZEH, PH, SEH, SEH, SEH, SEH, SEH, SEH, SEH, SE
CEC COM	NAME OCI DIRECTOR OF THE COLOR
SSIN TICK	REPSING TOO TOO TOO TOO TOO TOO TOO TOO TOO TO
SWI NA J NA J NA J HO SPR MER MER	NA 1 NA 1 FAC MER ME- SIO SENS SENS SENS SENS SENS SENS SENS SEN
CAT CAT CAT SALYN	S D D D D D D D D D D D D D D D D D D D
2834 2834 77.7.7 MANAN MONAN MONAN MANAN M	252C 252C 252C 252C 30S 30S 30S 30S 30S 30S 30S 30S 30S 30S
Human Gene SWISSNEW- ID:P28340 DNA POL YMERASE DELTA CATAL YTIC CHAIN (EC 2.7.7.7) - HOMO SAPIENS (HUMAN), 1107 aa.jpcis:SWISSPROT-ID:P28340 DNA POL YMERASE DELTA CATAL YTIC CHAIN (EC 2.7.7.7 - HOMO SAPIENS (HUMAN), 1107 aa.	Human Gene SWISSNEW- ID:P25205 DNA REPLICATION LICENSING FACTOR MCM3 (DNA POLYMERASE ALPHA HOLOENZYME-ASSOCIATED PROTEIN PI) (RLF BETA SUBUNIT) (PI02 PROTEIN) - HOMO SAPIENS (HUMAN), 808 aa. jpcis:SWISSPROT-ID:P25205 DNA REPLICATION LICENSING FACTOR MCM3 (DNA POLYMERASE ALPHA HOLOENZYME-ASSOCIATED PROTEIN PI) (RLF BETA SUBUNIT) (PI02 PROTEIN) - HOMO SAPIENS (HUMAN), 808 aa.
8	9
a a a a a a a a a a a a a a a a a a a	neta
polymerase	polymerase
NON- CONSER VATIVE	NON- CONSER VATIVE
NON- CONSI VATIN	NON- CONS VATIV
G	ြေ
Arg (785)	Gly (786)
0.	
g. T.	Arg
(2)	l _{cp}
<u>S</u>	
₹	0
SCEA SAG	CTCAGACCATGTC CTTCGGATGCAC[C/ G]GTTACAGAGCAC CTGGGGAGCAGGA
CAC	7777 6C¢ 6C¢ 6C¢ 6C¢
27.2 23.4 7.0 20.0 20.0 20.0	SAD
0.00 0.00 V	000 000 000 000 000 000 000 000 000 00
AGGTCCTCCTGA ATTGGGATGGCCIA /GJAGGTGCATCAT CATCATCCCAGAG G	CTCGGACC CTTCGGATC GJGTTACAG CTGGGGAG
3340	1593
214	858
1022	9958
cg44022214	cg43958858
	557
<u> </u>	

<u> </u>	<u>~</u>	<u> </u>	<u> </u>
12 (12p13)	12 (12p13)	12 (12p13)	(11q22)
0	0	0	2.406-59
Human Gene SWISSPROT- ID:P22460 VOLTAGE-GATED POTASSIUM CHANNEL PROTEIN KV1.5 (HK2) (HPCN1) - HOMO SAPIENS (HUMAN), 613 aa.	Human Gene SWISSPROT- ID:P22460 VOLTAGE-GATED POTASSIUM CHANNEL PROTEIN KVI.5 (HK2) (HPCNI) - HOMO SAPIENS (HUMAN), 613 aa.	Human Gene SWISSPROT- ID:P22460 VOLTAGE-GATED POTASSIUM CHANNEL PROTEIN KVI.5 (HK2) (HPCNI) - HOMO SAPIENS (HUMAN), 613 aa.	Human Gene Similar to SWISSPROT-ID:P50280 MATRIL YSIN PRECURSOR (EC 3.4.24.23) (PUMP-1 PROTEASE) (UTERINE METALLOPROTEINASE) (MATRIX METALLOPROTEINASE-7) (MMP-7) (MATRIN) - RATTUS NORVEGICUS (RAT), 267 aa.
potassium_chan nel	potassium_chan nel	potassium_chan nel	protease
NON- CONSER VATIVE	NON- CONSER VATIVE	NON- CONSER VATIVE	NON- CONSER VATIVE
Teu (787)	Gly (788)	Ala (789)	(790)
Gln	Arg	Pro	Asp
<u> </u>	0	უ	O
Α	U	U	_ව
CGCTTTGAGACGC AGCTGGGCACCCJA TJGGCGCAGTTCCC CAACACTCCTG	GGGGGACGAGGCC ATGGAGCGCTTC[C /G]GCGAGGATGAG GGCTTCATTAAAG A	CATTAAAGAAGAG GAGAAGCCCCTG[C /G]CCCGCAACGAG TTCCAGCGCCAGG	TGGAGGGGATGCT CATTTGATGAGG/ CJATGAAAGGTGG ACCAACAATTTCA G
641	898	910	868
cg42534568	cg42534568	cg42534568	cg43154190
558		260	561

11 (11922)	6 (spter)	
2.40E-59	1.60E-124 6	0
Human Gene Similar to SWISSPROT-ID:P50280 MATRIL YSIN PRECURSOR (EC 3.4.24.23) (PUMP-1 PROTEASE) (UTERINE METALLOPROTEINASE) (MATRIX METALLOPROTEINASE-7) (MATRIX) - (MATRIX) - RATTUS NORVEGICUS (RAT), 267 aa.	Human Gene Homologous to SWISSPROT-ID:P16083 NAD(P)H DE#YDROGENASE (QUINONE) 2 (EC 1.6.99.2) (QUINONE REDUCTASE) (DT- DIAPHORASE) (AZOREDUCTASE) (PHYLLOQUINONE REDUCTASE) (MEMADIONE REDUCTASE) + HOMO SAPIENS (HUMAN), 231 aa.	Human Gene TREMBLNEW- ID:G2725625 ACETOLACTATE SYNTHASE - HOMO SAPIENS (HUMAN), 632 aa.
protease	reductas e	synthase
NON- CONSER VATIVE	NON- CONSER VATIVE	NON- CONSER VATIVE
Thr (791)	8(7) (797)	Glu (793)
Arg	dg)	Gly
U	f-v	F
ტ	0	ပ
GATGAAAGGTGGA C CCAACAATTTCA[G /C]AGAGTACAACT TACATCGTGTTGCG	ATTCTACGATTCCG C GTTTGCTCCAG[G/T]GTAAACTAGCGCT CCTTTCCGTAAC	CGCTGCCTTCTCCC (GAAAGGTCTGC[C/ TJCCTTCACGCGTT CGGCTTCCCGCAG
923	694	1081
cg43154190	cg43927549	cg43325541
562	563	564

7.40E-65	7.40E-65
Human Gene Similar to SWISSNEW-ID:P39062 ACETYL-COENZYME A SYNTHETASE (EC 6.2.1.1) (ACETATE-COA LIGASE) (ACYL- ACTIVATING ENZYME) (ACETYL-COA SYNTHASE) - BACILLUS SVNTHASE) - BACILLUS SUBTILIS, 572 aa.[pcis:SWISSPROT-ID:P39062 ACETYL-COENZYME A SYNTHETASE (EC 6.2.1.1) (ACETATE-COA LIGASE) (ACYL- ACTIVATING ENZYME) (ACETYL-COA SYNTHASE) - BACILLUS SYNTHASE) - BACILLUS SYNTHASE) - BACILLUS	Human Gene Similar to SWISSNEW-ID:P39062 ACETYL-COENZYME A SYNTHETASE (EC 6.2.1.1) (ACETATE-COA LIGASE) (ACYL- ACTIVATING BNZYME) (ACETYL-COA SYNTHASE) - BACILLUS SUBTILIS, 572 aa. pcis:SWISSPROT-ID:P39062 ACETYL-COENZYME A SYNTHETASE (EC 6.2.1.1) (ACETATE-COA LIGASE) (ACYL- ACTIVATING ENZYME) (ACETYL-COA SYNTHASE) - BACILLUS SYNTHASE) - BACILLUS SYNTHASE) - BACILLUS
synthase	synthase
NON- CONSER VATIVE	NON- CONSER VATIVE
Leu (794)	Thr (795)
Ser	Ala
F	4
<u>U</u>	O .
GTGAAGGCATTTG TGGTCCTGGCCT[C/ TJGCAGTTCCTGTC CCATGACCCAGAA	GACTGTCACAGGG AAAATTCAACGA[G /A]CCAAGCTTCGA GACAAGGAGTGGA A
1474	1617
	cg43064068
565	999

2 (2p21)	2 (2p21)	8 (8p21)	5 (5q35.1)
0			8.30E-240
Human Gene SWISSPROT- ID:P23945 FOLLICLE STIMULATING HORMONE RECEPTOR PRECURSOR (FSH- R) (FOLLITROPIN RECEPTOR) - HOMO SAPIENS (HUMAN), 695 8a.	Human Gene SWISSPROT- ID:P23945 FOLLICLE STIMULATING HORMONE RECEPTOR PRECURSOR (FSH- R) (FOLLITROPIN RECEPTOR) - HOMO SAPIENS (HUMAN), 695 aa.	Human Gene SWISSPROT- ID:P35348 ALPHA-1A ADRENERGIC RECEPTOR (ALPHA 1A-ADRENOCEPTOR) (ALPHA-1C ADRENBERGIC RECEPTOR) - HOMO SAPIENS (HUMAN), 466 aa.	Human Gene SWISSPROT- ID:P21728 D(1A) DOPAMINE RECEPTOR - HOMO SAPIENS (HUMAN), 446 aa.
m7	tm.7	tm7	tm7
NON- CONSER VATIVE	NON- CONSER VATIVE	NON- CONSER VATIVE	NON- CONSER VATIVE
Ala (796)	71hr (797)	(798) (798)	Met (799)
Ţ.	Asn	Arg	91
ن ن	U	H	ຽ
∢	∢	ر ن	ပ
GCAAGAAGTTGAT TATATGACTCAG[A /G]CTAGGGGTCAG AGATCCTCTCTGGC	AAGGCCAACACC TGCTCTACATCA[A/ CJCCCTGAGGCCTT CCAGAACCTTCCC	GAATGTCTTGAGA ATCCAGTGTCTC[C/ TJGCAGAAAGCAGT CTTCCAAACATGC	TCTCTCTGGAGAA GATCCAACCCAT[C /G]ACACAAACGG TCAGCACCAACC T
1119	535	1475	1590
cg36988276	cg36988276	cg32296848	cg2524739
567			570

				<u>3 (3q25)</u>
8.50E-199	1.40E-196	2.50E-160	2.10E <i>-61</i>	2.205-207
Human Gene SWISSPROT- ID:P04001 GREEN-SENSITIVE OPSIN (GREEN CONE PHOTORECEPTOR PIGMENT) - HOMO SAPIENS (HUMAN), 364 aa.	Human Gene TREMBLNEW- ID:G2736282 G PROTEIN COUPLED RECEPTOR - HOMO SAPIENS (HUMAN), 362 aa.	Human Gene TREMBL/NEW- ID:E1246031 OLFACTORY RECEPTOR - HOMO SAPIENS (HUMAN), 312 aa.	Human Gene Similar to SWISSPROT-ID:P30975 TACHYKININ-LIKE PEPTIDES RECEPTOR 99D (DTKR) - DROSOPHILA MELANOGASTER (FRUIT FLY), 519 aa.	Human Gene SWISSPROT- ID:P26022 PENTAXIN- RELATED PROTEIN PIX3 PRECURSOR (TUMOR NECROSIS FACTOR- INDUCIBLE PROTEIN TSG-14) - HOMO SAPIENS (HUMAN), 381 88.
tm7	rm7	m7	Tm7	tnf
NON- CONSER VATIVE	NON- CONSER VATIVE	NON- CONSER VATIVE	NON- CONSER VATIVE	NON- CONSER VATIVE
Ile (800)	Gly (801)	Ser (802)	Thr (803)	(804)
Thr	Ser	Phe . ,	E	Met
H	O	υ	O	<u> </u>
O	∢	H	, E	V
AGTGTCTGGATGA TCTTTGTGGGTCA[C/ TJTGCATCCGTTTT CACAAATGGGCTT	CATCTTCTCCATCA ACCTCTTCAGC[A/G JGCATTTTCTTCCTC ACGTGCATGAG	TCTTTIGTGGACAT CTGCTTCTCCT[T/C] CACCACCGTCCCC AAGATGCTGGCC	GGCCCTGAGAGCA ACACCACGGGCA[T /C]CACAGCCTTCTC CATGCCCAGCTGG	TGGAAGCGTGCAT CCAGTGAGACCA[A MJTGAGGCTTGAGT CTTTTAGTGCCTG
394	519	285	89	
cg2320320	cg43264978	cg3003708		cg43336100
571	572	573	574	

∞		19 (19p13.3)	12
2.30E-55	0	4.30E-275	1.70E-177
Human Gene Similar to TREMBLNEW-ID:G2653845 TNF RECEPTOR-RELATED RECEPTOR FOR TRAIL - HOMO SAPIENS (HUMAN), 386 aa.	Human Gene SPTREMBL- ID:Q14872 METAL- REGULATORY TRANSCRIPTION FACTOR - HOMO SAPIENS (HUMAN), 753 aa.	Human Gene SWISSPROT- ID:P35269 TRANSCRIPTION INITIATION FACTOR IIF, ALPHA SUBUNIT (TFIIF- ALPHA) (TRANSCRIPTION ALPHA) (TRANSCRIPTION HOMO SAPIENS (HUMAN), 517 aa.	Human Gene SPTREMBL- ID:Q07279 TRANSCRIPTION FACTOR NF-E2 - MUS MUSCULUS (MOUSE), 373 aa.
tnfreceptor	transcriptfactor	transcriptfactor	transcriptfactor
NON- CONSER VATIVE	NON- CONSER VATIVE	NON- CONSER VATIVE	NON- CONSER VATIVE
Pro (805)	His (806)	Phe (807)	Leu (808)
Leu	Tyr	Leu	Phe
O	ڻ ت	Ę	Ö
<u></u>	V.	U	ပ
GAGGCGCGGGGAG T CCAGGCTGGGCT (CJCCGGGTCCCCA AGACCCTTGTGCTC	ACTCGCACGTGGA TCCTGAGGCTGT[A/ GJAGAGGTAAGGA AGGCTTTGCCACA G	CATTGACAGCGAG GCCTCCTCAGCC[C/ TJTCTTCATGGCGA AGAAGAAGACGCC	TGACAGAGCTGTA CCGTGACATTTT[C/ GJCAGCACCTTCGG GATGAATCAGGCA
234	2857	1285	1346
cg43335562	cg43140548	cg43011561	cg43998970
576		578	579

9 (9434)						_											 		
6.50E-192																			
Human Gene SWISSPROT-	[ID:P16442	FUCOSYLGLYCOPROTEIN	ALPHA-N-	ACETYLGALACTOSAMINYLT	RANSFERASE (EC 2.4.1.40)	(HISTO-BLOOD GROUP A	TRANSFERASE) (A	TRANSFERASE) /	FUCOSYLGLYCOPROTEIN 3-	ALPHA-	GALACTOSYLTRANSFERASE	(EC 2.4.1.37) (HISTO-BLOOD	GROUP B TRANSFERASE) (B	TRANSFERASE) (NAGAT) -	HOMO SAPIENS (HUMAN), 354	88.			
transferase																			
NON-	CONSER	VATIVE																	
	608)																 	 	
Pro																	 	 	
드			_														 	 	
<u>C</u>																			
CACTACTATGTCTT C	CACCGACCAGC[C/	TJGGCCGCGGTGCC	CCGCGTGACGCTG					•										 	
			_																
cg2537639 464	3																		
280																			

9 (9434)									_	-										
6.50E-192																				
Human Gene SWISSPROT-	ID:P10442	FUCOSYLGLYCOPROTEIN	ALPHA-N-	ACETYLGALACTOSAMINYLT	RANSFERASE (EC 2.4.1.40)	(HISTO-BLOOD GROUP A	TRANSFERASE) (A	TRANSFERASE) /	FUCOSYLGLYCOPROTEIN 3-	ALPHA-	GALACTOSYLTRANSFERASE	(EC 2.4.1.37) (HISTO-BLOOD	GROUP B TRANSFERASE) (B	TRANSFERASE) (NAGAT) -	HOMO SAPIENS (HUMAN), 354	aa.				
transferase																				
NON-	CONSEK	VATIVE												,			-			
Gly	(810)																			
Arg															_			-		
0																				
ပ															•					
TCGGCAGCTGTCA C	GTGCTGGAGGTG[C	/G]GCGCCTACAAG	CGCTGGCAGGACG										,					-		
1																				
581 cg2537639 523																				
581																				

9 (9434)																				
6.50E-192																			•	
Human Gene SWISSPROT-	(ID:P16442	FUCOSYLGLYCOPROTEIN	ALPHA-N-	ACETYLGALACTOSAMINYLT	RANSFERASE (EC 2.4.1.40)	(HISTO-BLOOD GROUP A	TRANSFERASE) (A	TRANSFERASE)/	FUCOSYLGLYCOPROTEIN 3-	ALPHA-	GALACTOSYLTRANSFERASE	(EC 2.4.1.37) (HISTO-BLOOD	GROUP B TRANSFERASE) (B	TRANSFERASE) (NAGAT) -	HOMO SAPIENS (HUMAN), 354	38.				
transferase							10 -								•					
NON-	CONSER	VATIVE																		-
Ile	(811)																			
Phe																				
⋖			_					_												
T														,				 		
GGTGTGCGTGGAC	GTGGACATGGAG[T	/A]TCCGCGACCAC	GTGGGCGTGGAGA	<u></u>													-			
																				
cg2537639 643																				
582																				

9 (9q34)	
6.50E-192	
Human Gene SWISSPROT- ID:P16442 FUCOSYLGLYCOPROTEIN ALPHA-N- ALPHA-N- RANSFERASE (EC.24.1.40) (HISTO-BLOOD GROUP A TRANSFERASE) (A TRANSFERASE) / FUCOSYLGLYCOPROTEIN 3- ALPHA- ALPHA- GALACTOSYLTRANSFERASE (GC.24.1.37) (HISTO-BLOOD GROUP B TRANSFERASE) (B TRANSFERASE) (NAGAT) - HOMO SAPIENS (HUMAN), 354 aa.	
transferase	
NON- CONSER VATIVE	
Ser (812)	
Gly	
∢ .	
<u>o</u>	
TCCGCTGTTCGGCA G CCCTGCACCC[G/ A]GCTTCTACGGAA GCAGCCGGGAGGC	
cg2537639 700	
583	

9 (9q34)
6.50E-192 9 (9q34)
Human Gene SWISSPROT- ID:P16442 FUCOSYLGLYCOPROTEIN ALPHA-N- ACETYLGALACTOSAMINYLT RANSFERASE (EC 24 1.40) (HISTO-BLOOD GROUP A TRANSFERASE) (A TRANSFERASE) / FUCOSYLGLYCOPROTEIN 3- ALPHA- GALACTOSYLTRANSFERASE (EC 24.1.37) (HISTO-BLOOD GROUP B TRANSFERASE) (B TRANSFERASE) (MAGAT) - HOMO SAPIENS (HUMAN), 354 aa.
transferase
NON- CONSER VATIVE
(813)
Leu
4
O
CAAGGACGAGGGC C GATITCTACTAC[C/ AJTGGGGGGGGTTCT TCGGGGGGGTCGGT
793
cg2537639
584

9 (9q34)	15	3 (3p25)
6.50E-192	0	0
Human Gene SWISSPROT- ID:P16442 FUCOSYLGL YCOPROTEIN ALPHA-N- ACETYLGALACTOSAMINYLT RANSFERASE (EC 2.4.1.40) (HISTO-BLOOD GROUP A TRANSFERASE) (A GALACTOSYLTRANSFERASE (EC 2.4.1.37) (HISTO-BLOOD GROUP B TRANSFERASE) (B TRANSFERASE) (NAGAT) - HOMO SAPIÈNS (HUMAN), 354 aa.	Human Gene SWISSPROT- ID:Q04671 P PROTEIN (MELANOCYTE-SPECIFIC TRANSPORTER PROTEIN) - HOMO SAPIENS (HUMAN), 838 aa.	Human Gene SWISSPROT- ID:P31641 SODIUM- AND CHLORIDE-DEPENDENT TAURINE TRANSPORTER - HOMO SAPIENS (HUMAN), 620 aa.
transferase	transport	transport
NON- CONSER VATIVE	NON- CONSER VATIVE	NON- CONSER VATIVE
(814)	Asp (815)	Met (816)
Val	Ala	Ile
₹	∢	២
<u>0</u>	<u>.</u> ပ_	U
GTTCTTCGGGGGG TCGGTGCAAGAG[G /A]TGCAGCGGCTC ACCAGGGCCTGCC A	CACATCGTGGTGG AGCTGACCCAGG[C AJTGACGCTTTGG GCTCCAGGTGGCG G	GTCTGAAAGATTT CCACAAGGACAT[C /G]CTGAAGCCCTC ACCAGGGAAGAGC C
	3249	427
	cg42742340	cg41653463
585	586	587

			T	
5 (5p15.3)	5 (5p15.3)		17	10 (10q25)
0	0	0	0	0
Human Gene SWISSPROT- ID:Q01959 SODIUM- DEPENDENT DOPAMINE TRANSPORTER (DA TRANSPORTER) (DAT) - HOMO SAPIENS (HUMAN), 620 aa.	Human Gene SWISSPROT- ID:Q01959 SODIUM- DEPENDENT DOPAMINE TRANSPORTER (DA TRANSPORTER) (DAT) - HOMO SAPIENS (HUMAN), 620 aa.	Human Gene SPTREMBL- ACC:Q16084 P130 - HOMO SAPIENS (HUMAN), 1139 aa.	Human Gene SWISSPROT- ACC:P42694 HYPOTHETICAL PROTEIN KIAA0054 - Homo sapiens (Human), 1942 aa.	Human Gene SWISSNEW- ACC:P46013 ANTIGEN KI-67 - Homo sapiens (Human), 3256 aa.
transport	transport	UNCLASSIFIE D	UNCLASSIFIE D	UNCLASSIFIE D
NON- CONSER VATIVE	NON- CONSER VATIVE	NON- CONSER VATIVE	NON- CONSER VATIVE	NON- CONSER VATIVE
His (817)	Ser (818)	Pro (819)	Arg (820)	Тър (821)
Asp	Phe	Ala	Giy	Leu
O	ပ	o	_ල	ڻ ا
5	<u> </u>	ا ا	U	-
CAAGTTCACCAAC AACTGCTACAGG[G /C]ACGCGATTGTC ACCACCTCCATCA A	TCCTCCGGCTTCGT CGTCTTCTCCT[T/C] CCTGGGGTACATG GCACAGAAGCAC	CTGCGGTAGCTGT CCCAGGCCTCGG[C /G]CCGCGCCCT CGTCCATGTTGAG G	GCATAGGACATGG CGGGCTTGCCCC[C/ G]CGCAGAGCTCTG GGGGCTACTGCTA	CAACCCCTAGAAG ACCTGGCTGGCT[7/ GJGAAAGAGCTCTT CCAGACACCAGTA
1165	1232	4776	522	4604
cg40351913	cg40351913	cg43955093	cg43055918	cg43968854
288	589		165	592

4 (4922)				
0	0	0	0	0
Human Gene SWISSPROT- ACC:P55157 MICROSOMAL TRIGL YCERIDE TRANSFER PROTEIN, LARGE SUBUNIT PRECURSOR - Homo sapiens (Human), 894 aa.	Human Gene SPTREMBL- ACC:Q15840 ZINC FINGER PROTEIN BASONUCLIN - HOMO SAPIENS (HUMAN), 994 aa.	Human Gene SPTREMBL- ACC:Q15840 ZINC FINGER PROTEIN BASONUCLIN - HOMO SAPIENS (HUMAN), 994 aa.	Human Gene SPTREMBL- ACC:Q15840 ZINC FINGER PROTEIN BASONUCLIN - HOMO SAPIENS (HUMAN), 994 aa.	Human Gene SPTREMBL- ACC:Q15840 ZINC FINGER PROTEIN BASONUCLIN - HOMO SAPIENS (HUMAN), 994 aa.
UNCLASSIFIE D	UNCLASSIFIE D	UNCLASSIFIE D	UNCLASSIFIE D	UNCLASSIFIE D
NON- CONSER VATIVE	NON- CONSER VATIVE	NON- CONSER VATIVE	NON- CONSER VATIVE	NON- CONSER VATIVE
Leu (822)	His (823)	Cys (824)	Tyr (825)	Ile (826)
Phe	ujo G	Ser	Asn	Asn
_ව	E-	H	E ·	H
<u>-</u>	5	¥	V	∀
CCATTGITCAAGA CATCCTACGITT[17/ GJGAAATGCCTGCA AGCAAAATTGTCC	ACAATTCAGAGAG GGAGACTGAGCA[GTJACACCAGCAT TGATCATGGTGCC AA	ATCAGGAAAGGTG , TTGGATCACTGG[A /T]GCATCATGACC AGTGAGGAAGAAG T	CCCCAAACAGGAA GTCCATGGGCCC[A AJACCCTGACAGC AGCTTCTTAACTTC	CCCAAACAGGAAG TCCATGGGCCCA[A /I]CCCTGACAGCA GCTTCTTAACTTCC
1841	2001	553	937	938
				cg43262121 5
593	594	595	965	597

	6 (6q14)			(cen)
	9) 9	6		2 (2cen)
0	9.1e-313	2.00E-301	6.10E-236	3.00E-227
Human Gene SWISSPROT- ACC:P02771 ALPHA- FETOPROTEIN PRECURSOR (ALPHA-FETOGLOBULIN) (ALPHA-1- FETOPROTEIN) - Homo sapiens (Human), 609 aa.	Human Gene SWISSPROT. ACC:P21589 5'-NUCLEOTIDASE PRECURSOR (EC 3.1.3.5) (ECTO-NUCLEOTIDASE) (5'- NT) (CD73 ANTIGEN) - Homo sapiens (Human), 574 aa.	Human Gene SWISSNEW- ACC:P42568 AF-9 PROTEIN - Homo sapiens (Human), 568 aa.	Human Gene SWISSNEW- ACC:043913 ORIGIN RECOGNITION COMPLEX SUBUNIT 5 - Homo sapiens (Human), 435 aa.	Human Gene SWISSPROT. ACC:P09529 INHIBIN BETA B CHAIN PRECURSOR (ACTIVIN BETA-B CHAIN) - Homo sapiens (Human), 407 aa.
UNCLASSIFIE D	UNCLASSIFIE D	UNCLASSIFIE D	UNCLASSIFIE D	UNCLASSIFIE D
NON- CONSER VATIVE	NON- CONSER VATIVE	NON- CONSER VATIVE	NON- CONSER VATIVE	NON- CONSER VATIVE
(827)	Thr (828)	Ser (829)	Arg (830)	Ala (831)
Glu	Ala	Ala	Gly	Ser
O	∢	L	¥.	ပ
₹ .	<u>.</u>	ຍ	ن	•
CTGGAAGAACTTT GCCATGAGAAAG[A/G]AATTTTGGAG AAGTACGGACATT CA	AATGATTAACAAC AACCTGAGACAC[G /A]CGGATGAAATG TTCTGGAACCACG T	GCCCCAGGCATG GCTAGCTCGTGT[G/ TJCCGTGCAGGTGA AGCTGGAGCTGGG	CAGCTTTCCATCCA TTTTTATTTAT[G/A] GACATACTGCTAG TGGAAAGACCTA	CAGGTGTCCTGCG AGCCACCCGGGG[ACJTCCGGTGGC GGGGGTGGCGGGG GC
501	1235	367		3034
	cg44928804			cg42913861
865	665	009	601	602

15		12
1.40E-188	4.20E-150	3.70E-142
UNCLASSIFIE Human Gene SWISSPROT- D ACC:P09471 GUANINE NUCLEOTIDE-BINDING PROTEIN G(0), ALPHA SUBUNIT 1 - Homo sapiens (Human), 353 aa.	Human Gene Homologous to SWISSPROT-ACC:P14207 FOLATE RECEPTOR BETA PRECURSOR (FR-BETA) (FOLATE RECEPTOR 2) (FOLATE RECEPTOR, FETAL/PLACENTAL) (FLACENTAL) (FLACENTAL) (FLACENTAL) (FLACENTAL) (FLACENTAL) HOMO sapiens (Human), 255 aa.	Human Gene Homologous to SWISSPROT-ACC.P21583 STEM CELL FACTOR PRECURSOR (SCF) (MAST/CELL GROWTH FACTOR) (MGF) (C-KIT LIGAND) - Homo sapiens (Human), 273 ag.
UNCLASSIFIE D	UNCLASSIFIE D	UNCLASSIFIE D
NON- CONSER VATIVE	NON- CONSER VATIVE	NON- CONSER VATIVE
Gly (832)	Arg (833)	Gly (834)
હ્ય	Ser	Arg
ರ	O	_ව
∢	¥	V
AGAGGAGAGAGCC A GCCCTCGAGCGG[A /G]GCAAGGCGATT GAGAAAACCTCA A	GCCAGAGTTGCAG CATCAGGGCCAG[A /C]CTGAGCAGGAG ACCCCCAGTCCCA T	TAGGAATGACAGC , AGTAGCAGTAAT[A /GJGGAAGGCCAAA AATCCCCCTGGAG A
526	335	787
cg43249389	cg43919239	cg41642952
603	604	509

	10	9	m
1.60E-134	1.60E-116	2.20E-98	2.90E-87
Human Gene Homologous to SWISSNEW-ACC:P08637 LOW AFFINITY IMMUNOGLOBULIN GAMMA FC RECEPTOR III-1 PRECURSOR (FC-GAMMA RIII) (FCRIII) (IGG FC RECEPTOR III-1) (FC-GAMMA RIII-ALPHA) (CD16) (FCR-10) - Homo sapiens (Human), 254 aa.	Human Gene Homologous to SWISSPROT-ACC:P50539 MAX INTERACTING PROTEIN 1 (MXII PROTEIN) - Homo sapiens (Human), 228 aa.	Human Gene Similar to TREMBLNEW-ACC:AAD38008 GLYOXALASE-I (EC 4.4.1.5) - HOMO SAPIENS (HUMAN), 184 aa.	Human Gene Similar to SWISSPROT-ACC:P49913 ANTIBACTERIAL PROTEIN FALL-39 PRECURSOR (FALL-39 PEPTIDE ANTIBIOTIC) (ANTIMICROBIAL PROTEIN CAP-18) (LL-37) - Homo sapiens (Human), 170 aa.
UNCLASSIFIE D	UNCLASSIFIE D	UNCLASSIFIE D	UNCLASSIFIE D
NON- CONSER VATIVE	NON- CONSER VATIVE	NON- CONSER VATIVE	NON- CONSER VATIVE
(835)	??? (836)	Ala (837)	(838)
Arg	Set	ng G	Thr
U	ω	9	V .
0	ပ	L	ပ
TGTTCCTGGAGCCT CAATGGTACAG[G/ CJGTGCTCGAGAAG GACAGTGTGACTC	GGGCACAGAACA CAGCAGCGGAG[C/S]AGCAACACCA GCACTGCCAACAG AT	ATTGCCATTGTGGT AACTCTGGGTC[T/G]CATCATCTTCAGT GCCCCAATTGTG	GTGAAGCGGTGTA TGGGACAGTGA[C /A]CCTCAACCAGG CCAGGGGCTCCTTT
221	391	6091	521
	2		cg42556108 5
909	607	809	609

·	4			14 (14q11.2)
2.30E-85	2.80E-73	5.90E-64	5.90E-64	1.00E-59
Human Gene Similar to SWISSPROT-ACC:P01282 VASOACTIVE INTESTINAL PEPTIDE PRECURSOR (VIP) - Homo sapiens (Human), 170 aa.	Human Gene Similar to SPTREMBL-ACC:Q94218 CODED FOR BY C. ELEGANS CDNA CM10H5 - CAENORHABDITIS ELEGANS, 589 aa.	Human Gene Similar to SPTREMBL-ACC:076087 GAGE-8 - HOMO SAPIENS (HUMAN), 117 aa.	Human Gene Similar to SPTREMBL-ACC:076087 GAGE-8 - HOMO SAPIENS (HUMAN), 117 aa.	Human Gene Similar to REMTREMBL-ACC:G238693 T CELL RECEPTOR VARIABLE ALPHA CHAIN - HOMO SAPIENS (HUMAN), 143 aa (fragment).
UNCLASSIFIE D	UNCLASSIFIE D	UNCLASSIFIE D	UNCLASSIFIE D	UNCLASSIFIE D
NON- CONSER VATIVE	NON- CONSER VATIVE	NON- CONSER VATIVE	NON- CONSER VATIVE	NON- CONSER VATIVE
Pro (839)	Phe (840)	Val (841)	Val (842)	Arg (843)
Oln	Val	Met	Met	Gly
O	E-	ව	ົນ	O
<	ပ	«	∀	ຽ
AGTGACTTCAGTA AACTCTTGGGTC[A/ CJACTTTCTGCCAA AAAGTACCTTGAG	CGGTATAACGTCA AAAATCCTGTTT[G/ TJTCAGCCAAGGTT CAGAAATTGCCTC	AAGGCGCTATGTA CAGCCTCCTGAA[A /GJTGATTGGGCCT ATGCGGCCCGAGC A	TGAAGATGGTCCT GATGGCAGGAG[A/G]TGGACCCGCC AAATCCAGAGGAG GT	ATTACTGAAGGGT GGAGAACAGAAG[G/CJGTCATGAAAA AATATCTGCTTCAT T
487	1052	283	505	260
cg36842490	cg43942549	cg42381630	cg42381630	cg3004395
610	119	612	613	614

1.20E-58	1.60E-54	1.60E-54	1.60E-54	1.60E-54
Human Gene Similar to SWISSNEW-ACC:076070 GAMMA-SYNUCLEIN (PERSYN) (BREAST CANCER- SPECIFIC GENE 1 PROTEIN) - Homo sapiens (Human), 127 aa.	Human Gene Similar to REMTREMBL-ACC:G33509 T CELL RECEPTOR - HOMO SAPIENS (HUMAN), 118 aa (fragment).	Human Gene Similar to REMTREMBL-ACC:G33509 T CELL RECEPTOR - HOMO SAPIENS (HUMAN), 118 aa (fragment).	Human Gene Similar to REMTREMBL-ACC:G33509 T CELL RECEPTOR - HOMO SAPIENS (HUMAN), 118 aa (fragment).	Human Gene Similar to REMTREMBL-ACC:G33509 T CELL RECEPTOR - HOMO SAPIENS (HUMAN), 118 aa (fragment).
UNCLASSIFIE D	UNCLASSIFIE D	UNCLASSIFIE D	UNCLASSIFIE D	UNCLASSIFIE D
NON- CONSER VATIVE	NON- CONSER VATIVE	NON- CONSER VATIVE	NON- CONSER VATIVE	NON- CONSER VATIVE
Glu (844)	Gln (845)	Met (846)	Gln (847)	Ser (848)
Val	Leu	Val	Leu	Phe
<u>F</u>	∢	∢	V	ပ
V	 	හ	Ŀ →	[
CACTTCCTCTTCT CTTTGGATGCC[AT JCACCCTCTGTTG GGGGCAGATGG	GAAGACAAGGTGG TACAAAGCCCTC[T/ AJATCTCTGGTTGT CCACGAGGGAGAC	TGTAACTCTCAATT GCAGTTATGAA[G/ AJTGACTAACTTTC GAAGCCTACTATG	GAAGTGACTAACT TTCGAAGCCTAC[T/ A]ATGGTACAAGCA GGAAAAGAAAGCT	AGCATATTAGATA AGAAAGAACTTT[T /C]CAGCATCCTGA ACATCACAGCCAC C
733	289	342	364	475
cg43960645	cg2526759	cg2526759		c <u>g</u> 2526759
615	616	617	819	619

17 (17421.3 2)	17 (17q21.3 2)	
0	0	1.80E-157
Human Gene SWISSPROT- ID:P08514 PLATELET MEMBRANE GLYCOPROTEIN IIB PRECURSOR (GPIIB) (INTEGRIN ALPHA- IIB) (CD41) - HOMO SAPIENS (HUMAN), 1039 aa.	Human Gene SWISSPROT- ID:P08514 PLATELET MEMBRANE GI. YCOPROTEIN IIB PRECURSOR (GPIIB) (INTEGRIN ALPHA- IIB) (CD41) - HOMO SAPIENS (HUMAN), 1039 aa.	Human Gene SWISSNEW- DI:Q08722 LEUKOCYTE SURFACE ANTIGEN CD47 PRECURSOR (ANTIGENIC SURFACE DETERMINANT PROTEIN OA3) (INTEGRIN ASSOCIATED PROTEIN) (IAP) (MER6) - HOMO SAPIENS (HUMAN), 323 aa.[pcis:SWISSPROT-ID:Q08722 LEUKOCYTE SURFACE ANTIGEN CD47 PRECURSOR (ANTIGEN CD47 PRECURSOR (ANTIGENIC SURFACE DETERMINANT PROTEIN OA3) (INTEGRIN ASSOCIATED PROTEIN) (IAP) (MER6) - HOMO SAPIENS (HUMAN), 323 aa.
cadherin	cadherin	cadherin
FRAMES cadherin	FRAMES HIFT	FRAMES cadherin
His (849)	His (850)	(851) (851)
TET.	Pro	Arg
O	₹	de 8
gap	gap	<u>F</u>
TACAGAATATGTC GTCGGTGCCCCC[ga p/C]ACTTGGAGCTG GACCCTGGGAGCG G	GTCGGCTTCTTCAA GCGGAACCGGC[ga p/A]CACCCCTGGAA GAAGATGATGAAG A	GTCCATCACTTCAC TTCAGTTATTC[T/ga p]CCTAGGAGGTTG TATAGTCTTCTGA
1067	3285	2521
	cg40310734	cg43956660
	129	622

2	17	17	11 (11q23)	15 (15q15)
0	0	7 7 7 7	1.80E-203	0
Human Gene SWISSPROT- D:P12111 COLLAGEN ALPHA 3(VI) CHAIN PRECURSOR - HOMO SAPIENS (HUMAN), 3176 aa.	Human Gene SPTREMBL- ID:Q92804 PUTATIVE RNA BINDING PROTEIN RBP56 - HOMO SAPIENS (HUMAN), 592 aa.	Human Gene SPTREMBL- ID:Q92804 PUTATIVE RNA BINDING PROTEIN RBP56 - HOMO SAPIENS (HUMAN), 592 aa.	Human Gene SWISSNEW- ID:P06727 APOLIPOPROTEIN A- IV PRECURSOR (APO-AIV) - HOMO SAPIENS (HUMAN), 396 aa-[pcis:SWISSPROT-ID:P06727 APOLIPOPROTEIN A-IV PRECURSOR (APO-AIV) - HOMO SAPIENS (HUMAN), 396 aa.	Human Gene SWISSPROT- ID:P16452 ERYTHROCYTE MEMBRANE PROTEIN BAND 4.2 (P4.2) (PALLIDIN) - HOMO SAPIENS (HUMAN), 690 aa.
collagen	dna_ma_bind	dna_ma_bind	ųdo	glycoprotein
FRAMES collagen HIFT	FRAMES HIFT	FRAMES HIFT	FRAMES HIFT	FRAMES HIFT
Gly (852)	Arg (853)	Leu (854)	Asp (855)	Asn (856)
Glý	Arg	T _y r	Thr	Thr
O	_ව	U	O	4
dag	gab	deg	de 8	dead
CTCCAGGGATAGT TGGACAGAAGGG[g ap/G]AGACCCTGGC TACCCAGGACCAG CT	GCTATGGAGGCAA AATGGGAGGAAG[g ap/G]AAACGACTAC AGAAATGATCAGC GC	CGGTTACTCCAGTT ATGGACAAAGT[ga p/C]TATTCACAGTC CTATGGTGGTTATG	GGCCGAGCAGCTG CGGCGCCAGCTG[g ap/G]ACCCCCTACG CACAGCGCATGGA GA	CAGACTTCCACAG
2429	1837		584	1553
cg43970982	cg42175288	cg42175288		cg43065549
623	624	625	626	627

14 (14q11.2)	7	9	20
9.90E-70	1.20E-224	1.40E-261	9.60E-262
Human Gene Similar to SWISSPROT-ID:P16452 ERYTHROCYTE MEMBRANE PROTEIN BAND 4.2 (P4.2) (PALLIDIN) - HOMO SAPIENS (HUMAN), 690 aa.	Human Gene SWISSPROT- ID:PS0219 HOMEOBOX PROTEIN HB9 - HOMO SAPIENS (HUMAN), 401 aa.	Human Gene SWISSPROT- ID:P15260 INTERFERON- GAMMA RECEPTOR ALPHA CHAIN PRECURSOR (CDW119) - HOMO SAPIENS (HUMAN), 489 aa.	Human Gene SPTREMBL- ID:Q15802 SERINE/THREONINE PROTEIN KINASE KRS-2 - HOMO SAPIENS (HUMAN), 487 aa.
FRAMES glycoprotein HIFT	homeobox	interferon	kinase
FRAMES HIFT	FRAMES HIFT	FRAMES HIFT	FRAMES HIFT
(857)	Arg (858)	End (859)	His (860)
Leu	Pro	Leu	Te
U	ပ	gap	O
gap	gap	• •	gap
TGACCACGGGGTG CTGGATGCCTGC[ga P/C]TTATACATCCT GGACCGGCGGGGG A	GCGCCGCGAGACA AGGCAGCGGAC[g ap/G]CGCCTGCGGA CTTGAGGGACAGT GA	ATAAGTTACAATG CTTTTTTTGTTT[A/g ap]AAAAAAAAAA AAGTCTGTACTTTA	CTGTGGGGCTGGT TCTGTATCTGAT[ga p/CJATCATTCGATT ACGAAATAAAACG T
666		364	379
	cg41637704		cg43072541
628	629	630	631

13)			ନ୍ଥ
2 (2413)	61	61	1 (1p33)
2.40E-82	9.70E-214	9.70E-214	0
Human Gene Similar to SWISSPROT-ID:P25155 COAGULATION FACTOR X PRECURSOR (EC 3.421.6) (STUART FACTOR) (VIRUS ACTIVATING PROTEASE) (VAP) - GALLUS GALLUS (CHICKEN), 475 aa.	Human Gene SWISSPROT- ID:P01137 TRANSFORMING GROWTH FACTOR BETA 1 PRECURSOR (TGF-BETA 1) - HOMO SAPIENS (HUMAN), 390 aa.	Human Gene SWISSPROT- ID:P01137 TRANSFORMING GROWTH FACTOR BETA 1 PRECURSOR (TGF-BETA 1) - HOMO SAPIENS (HUMAN), 390 aa.	Human Gene SWISSPROT- ID:Q03167 TGF-BETA RECEPTOR TYPE III PRECURSOR (TGFR-3) (BETAGLYCAN) - HOMO SAPIENS (HUMAN), 849 aa.
protease	Îgi	J 81	tgfreceptor
FRAMES protease	FRAMES HIFT	FRAMES HIFT	FRAMES HIFT
(861)	Ala (862)	(863)	Ala (864)
His	Leu	Leu	Ala
<u></u>	_ව	ڻ ت	υ
	gap	бар	du 8
GTCAGCCGCTACC FOR TCGACCIGA PATATGGGCACATC AGAGACAAGGAAG C	CCGGGCAGAGCTG CGTCTGCTGAGG[g ap/G]CTCAAGTTAA AAGTGGAGCAGCA CG	CCGGGCAGAGCTG CGTCTGCTGAGG[g ap/G]CTCAAGTTAA AAGTGGAGCAGCA CG	AATCTCCGCACTG CAGGCCAGGGC[g ap/CJTGGCCAGGTA CAGAGAGGTCA CAGAGAGAGGTCA CA
1536	1317	1317	847
cg44032168	cg43931248	cg43931248	cg43272560
632	633	634	635

<u>v</u>	
5.20E-254	.406-182
5.20E	1.40E
DE DE AP- AP- (AN),	Human Gene SWISSPROT- ID:P53611 GERANYLGERANYL TRANSFERASE TYPE II BETA SUBUNIT (EC 2.5.1) (RAB GERANYLGERANYLTRANSFE GERANYL- GERANYL- GERANYL- GERANYL- GERANYL- GERANYL- GERANYL- GHUMAN, 331 aa. (HUMAN), 331 aa.
EDUN CAALL 24	THE SOR COMPOS
tm.7	FRAMES transferase
ES	TES TEST
FRAMES un7	FRAN
(865) 1 (865) 1	Pro (866)
Giy	Gin
E-	೮
des	des
CCAGGATCCATTTT gap GAGGATTATGG[gap //JGTGCTGGGACA CCATCAACTCCTCA	CAAATCCCCCGGTT E TCTTCATCTTG[gap/ GJACATGCTAAAAT GAAATTACGCAGT
İ	625
cg43266471 1067	c <u>8</u> 43995237
636	637

part	×	 	-
1.40E-182	6.40E-257	3.80E-252	3.80E-252
Human Gene SWISSPROT- ID:P53611 GERANYL GERANYL TRANSFERASE TYPE II BETA SUBUNIT (EC 2.5.1) (RAB GERANYLGERANYLTRANSFE RASE BETA SUBUNIT) (RAB GERANYL- GERANYLTRANSFERASE BETA SUBUNIT) (RAB GERANYLTRANSFERASE BETA SUBUNIT) (RAB GGTASE) - HOMO SAPIENS (HUMAN), 331 aa.	Human Gene SWISSPROT- ACC:P78539 SUSHI REPEAT- CONTAINING PROTEIN SRPX PRECURSOR - Homo sapiens (Human), 464 aa.	Human Gene SWISSNEW- ACC.Q13228 SELENIUM- BINDING PROTEIN 1 - Homo sapiens (Human), 472 aa.	Human Gene SWISSNEW- ACC:Q13228 SELENIUM- BINDING PROTEIN 1 - Homo sapiens (Human), 472 aa.
FRAMES transferase HIFT	UNCLASSIFIE D	UNCLASSIFIE D	UNCLASSIFIE D
FRAMES HIFT	FRAMES HIFT	FRAMES	FRAMES HIFT
Phe (867)	Arg (868)	Leu (869)	Gly (870)
Leu	Arg	Pro	Gly
O	O	gap	gap
ga G.	deg	o	9
TTCTTCATCTTGAC ATGCTAAAATG[gap /G]AAATTACGCAG TTTCTCTCTATCAA	CCGCCTCTGCTGCTGCT GCTGCTGCTGC[gap /G]GCGTCCCGCCC AGCGCAGCTTCC C	ATCCAGGCTGAGC TGGATCATCTGAGG /gapJGGCCTCCAGC CACCCGTTTTCCCT T	CCAGGCTGAGGTG GATCATCTGAGGIG /gap CCTCCAGCCA CCCGTTTTCCCTTG A
638	267	965	199
	cg43254094		cg44034555 c
638	639	640	641

	1 (1p36.2)	9 (9q22.2)	20	20
1.00E-251	1.00E-201	3.50E-178	3.20E-147	3.20E-147
Human Gene SWISSPROT- ACC:P18428 LIPOPOLYSACCHARIDE- BINDING PROTEIN PRECURSOR (LBP) - Homo sapiens (Human), 481 aa.	Human Gene SWISSPROT- ACC:P18615 RD PROTEIN - Homo sapiens (Human), 380 aa.	Human Gene SWISSPROT- ACC:P09467 FRUCTOSE-1,6- BISPHOSPHATASE (EC 3.1.3.11) (D-FRUCTOSE-1,6- BISPHOSPHATE 1- PHOSPHOHYDROLASE) (FBPASE) - Homo sapiens (Human), 337 aa.	Human Gene Homologous to SPTREMBL-ACC:Q15182 SNRNP POL YPEPTIDE B - HOMO SAPIENS (HUMAN), 285 aa.	Human Gene Homologous to SPTREMBL-ACC:Q15182 SNRNP POLYPEPTIDE B - HOMO SAPIENS (HUMAN), 285 aa.
UNCLASSIFIE D	UNCLASSIFIE D	UNCLASSIFIE D	UNCLASSIFIE D	UNCLASSIFIE D
FRAMES HIFT	FRAMES HIFT	FRAMES HIFT	FRAMES	FRAMES
Gly (871)	Asp (872)	(873)	(874)	Phe (875)
Val	Asp ·	Oly Oly	Pro	Phe
Ö	U	drag drag	deg	gap
gap	gap	o	ပ	9
AGCGAGTCCTCCG GGAGGCCCACAG[g ap/GJTTACTGCCTC CAGCTGCAGCAGT GA	CGTTCCAGAGGAG CATATCTGCTGA[ga p/C]TGATGACCTGC AAGAGTCATCCAG A	GGAACTCGAGCAC GTCGTCGGGGGA[C /gap]CCCAAGATCA CCGGCGCCCTCTG GT	ATTCCCGGGGGAG GGGCCCTGTAA[G /gap]GGAAACCAGA CAATCCCATGAGA CT	TCCCGGGGGAGGG GGCCCTGTAAGG[G /gsp]AAACCAGACA ATCCCATGAGACT CC
882	379	306	195	197
cg39711096	cg44128902	cg43946951	cg43948890	cg43948890
642	643	644	645	646

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5 (5q23)		vo	v
1.00E-107	2.50E-72	5.80E-50	5.80E-50
Human Gene Homologous to SWISSNEW-ACC:Q99075 HEPARIN-BINDING EGF-LIKE GROWTH FACTOR PRECURSOR (HB-EGF) (HBEGF) (DIPHTERIA TOXIN RECEPTOR) (DT-R) - Homo sapiens (Human), 208 aa.	Human Gene Similar to SPTREMBL-ACC:060869 EDF-1 PROTEIN - HOMO SAPIENS (HUMAN), 148 aa.	Human Gene Similar to TREMBLNEW-ACC: AAD38944 NJAC PROTEIN - HOMO SAPIENS (HUMAN), 99 aa.	Human Gene Similar to TREMBLNEW-ACC:AAD38944 NJAC PROTEIN - HOMO SAPIENS (HUMAN), 99 aa.
UNCLASSIFIE D	UNCLASSIFIE D	UNCLASSIFIE D	UNCLASSIFIE D
FRAMES HIFT	FRAMES HIFT	FRAMES HIFT	FRAMES HIFT
(877)	Gly (878)	Asp (879)	Asp (880)
Asp	Gl _y	Gly	Gly
ڻ ت	gap	gap	gap
gap	U	O	9
CTCTCGGCACTGGT GACTGGCGAGA[ga p/G]CCTGGAGCGG CTTCGGAGAGGGC TA	TCGTGGCCAGGTC CTTCTGCGTAAG[C/ gap]CCCTTGCTCTG CCGACCTTGCTGG	GGTCCAAATGCAA GTGCTCCCGGAA[G /gap]GGACCCAAGA TCCGCTACAGCGA CG	TCCAAATGCAAGT GCTCCCGGAAGG[G /gap]ACCCAAGATC CGCTACAGCGACG TG
373	681	450	452
			cg44010855 4
648	649	059	651
	eg43942004 373 CTCTCGGCACTGGT gap G Asp Glu FRAMES UNCLASSIFIE Human Gene Homologous to 1.00E-107 GACTGGCGAGA[ga (877) HIFT D SWISSNEW-ACC:Q99075 I.00E-107 PAGCTGGAGCGG CTTCGGAGCGG CTTCGGAGAGGGC REPARIN-BINDING EGF-LIKE I.00E-107 TA TA RECURSOR (HB-EGF) (HBEGT) RECURSOR (HB-EGF) (HBEGT) (DIPHTRIA TOXIN RECEPTOR) (DT-R) - Homo Sapiens (Human), 208 aa.	cg43942004 373 CTCTCGGCACTGGT gap G Asp Glu FRAMES UNCLASSIFIE Human Gene Homologous to 1.00E-107 CACTGGCGAGA[ga (877) HIFT D SWISSNEW-ACC;Q99075 1.00E-107 CACTGGCGAGA[ga (877) HIFT D SWISSNEW-ACC;Q99075 1.00E-107 CACTGGAGAGAGG TA HIFT D SWISSNEW-ACC;Q99075 1.00E-107 CACTGGAGAGAGG TA HIFT D SWISSNEW-ACC;Q99075 1.00E-107 CACTGGAGAGGG TA HIFT D SWISSNEW-ACC;Q99075 1.00E-107 CACTGGAGAGGG GB GB GB GB GB GB CACTGGAGAGGG GB GB FRAMES UNCLASSIFIE Human Gene Similar to 2.50E-72 GCGACCTTGCTCG GB GB GB FRAMES HIFT D SPTREMBL-ACC:060869 EDF-1 A A A A A HIFT D FRAMES	B cg43942004 373 CTCTGGGCACTGGT gap G Asp G G FRAMES UNCLASSIFIE Human Gene Homologous to GATCGGAGGGG G G G G G G G G

CLAIMS

WHAT IS CLAIMED IS:

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- 1. An isolated polynucleotide selected from the group consisting of:
 - a) a nucleotide sequence comprising one or more polymorphic sequences
 (SEQ ID NOS:1 651);
 - b) a fragment of said nucleotide sequence, provided that the fragment includes a polymorphic site in said polymorphic sequence;
 - c) a complementary nucleotide sequence comprising a sequence complementary to one or more of said polymorphic sequences (SEQ ID NOS:1 - 651); and
 - d) a fragment of said complementary nucleotide sequence, provided that the fragment includes a polymorphic site in said polymorphic sequence.
- 2. The polynucleotide of claim 1, wherein said polynucleotide sequence is DNA.
- 3. The polynucleotide of claim 1, wherein said polynucleotide sequence is RNA.
- 4. The polynucleotide of claim 1, wherein said polynucleotide sequence is between about 10 and about 100 nucleotides in length.
- 5. The polynucleotide of claim 1, wherein said polynucleotide sequence is between about 10 and about 90 nucleotides in length.

6. The polynucleotide of claim 1, wherein said polynucleotide sequence is between about 10 and about 75 nucleotides in length.

- 7. The polynucleotide of claim 1, wherein said polynucleotide is between about 10 and about 50 bases in length.
 - 8. The polynucleotide of claim 1, wherein said polynucleotide is between about 10 and about 40 bases in length.
- The polynucleotide of claim 1, wherein said polynucleotide is derived from a nucleic acid encoding a polypeptide related to angiopoietin, 4-hydroxybutyrate dehydrogenase, ATP-dependent RNA helicase, MHC Class I histocompatibility antigen, or phosphoglycerate kinase.
- 15 10. The polynucleotide of claim 1, wherein said polymorphic site includes a nucleotide other than the nucleotide listed in Table 1, column 5 for said polymorphic sequence.
- The polynucleotide of claim 1, wherein the complement of said polymorphic site includes a nucleotide other than the complement of the nucleotide listed in Table 1, column 5 for the complement of said polymorphic sequence.
 - 12. The polynucleotide of claim 1, wherein said polymorphic site includes the nucleotide listed in Table 1, column 6 for said polymorphic sequence.

13. The polynucleotide of claim 1, wherein the complement of said polymorphic site includes the complement of the nucleotide listed in Table 1, column 6 for said polymorphic sequence.

An isolated allele-specific oligonucleotide that hybridizes to a first polynucleotide at a polymorphic site encompassed therein, wherein the first polynucleotide is chosen from the group consisting of:

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(-)

- a) a nucleotide sequence comprising one or more polymorphic sequences
 (SEQ ID NOS:1 651) provided that the polymorphic sequence includes a nucleotide other than the nucleotide recited in Table 1, column 5 for said polymorphic sequence;
- b) a nucleotide sequence that is a fragment of said polymorphic sequence,
 provided that the fragment includes a polymorphic site in said polymorphic sequence;
- c) a complementary nucleotide sequence comprising a sequence complementary to one or more polymorphic sequences (SEQ ID NOS:1 -651), provided that the complementary nucleotide sequence includes a nucleotide other than the complement of the nucleotide recited in Table 1, column 5; and
- d) a nucleotide sequence that is a fragment of said complementary sequence, provided that the fragment includes a polymorphic site in said polymorphic sequence.
- The oligonucleotide of claim 14, wherein the oligonucleotide does not hybridize under stringent conditions to a second polynucleotide selected from the group consisting of:

a) a nucleotide sequence comprising one or more polymorphic sequences
 (SEQ ID NOS:1 - 651), wherein said polymorphic sequence includes the nucleotide listed in Table 1, column 5 for said polymorphic sequence;

b) a nucleotide sequence that is a fragment of any of said nucleotide sequences;

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- c) a complementary nucleotide sequence comprising a sequence complementary to one or more polymorphic sequences (SEQ ID NOS:1 -651), wherein said polymorphic sequence includes the complement of the nucleotide listed in Table 1, column 5; and
- d) a nucleotide sequence that is a fragment of said complementary sequence, provided that the fragment includes a polymorphic site in said polymorphic sequence.
- 16. The oligonucleotide of claim 15, wherein the oligonucleotide is between about 10 and about 51 bases in length.
 - 17. The oligonucleotide of claim 15, wherein the oligonucleotide identifies a polypeptide related to angiopoietin, 4-hydroxybutyrate dehydrogenase, ATP-dependent RNA helicase, MHC Class I histocompatibility antigen, or phosphoglycerate kinase.
 - 18. The oligonucleotide of claim 15, wherein the oligonucleotide is between about 15 and about 30 bases in length.
 - 19. A method of detecting a polymorphic site in a nucleic acid, the method comprising:
- a) contacting said nucleic acid with an oligonucleotide that hybridizes to a polymorphic sequence selected from the group consisting of SEQ ID NOS:
 1 651, or its complement, provided that the polymorphic sequence

includes a nucleotide other than the nucleotide recited in Table 1, column 5 for said polymorphic sequence, or the complement includes a nucleotide other than the complement of the nucleotide recited in Table 1, column 5; and

- b) determining whether said nucleic acid and said oligonucleotide hybridize; whereby hybridization of said oligonucleotide to said nucleic acid sequence indicates the presence of the polymorphic site in said nucleic acid.
- 20. The method of claim 19, wherein said oligonucleotide does not hybridize to said

 polymorphic sequence when said polymorphic sequence includes the nucleotide

 recited in Table 1, column 5 for said polymorphic sequence, or when the complement

 of the polymorphic sequence includes the complement of the nucleotide recited in

 Table 1, column 5 for said polymorphic sequence.
- The method of claim 19, wherein said oligonucleotide identifies a polypeptide related to angiopoietin, 4-hydroxybutyrate dehydrogenase, ATP-dependent RNA helicase, MHC Class I histocompatibility antigen, or phosphoglycerate kinase.
- The method of claim 19, wherein said oligonucleotide is between about 15 and about
 30 bases in length.
 - 23. A method of detecting the presence of a sequence polymorphism in a subject, the method comprising:
 - a) providing a nucleic acid from said subject;

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b) contacting said nucleic acid with an oligonucleotide that hybridizes to a
polymorphic sequence selected from the group consisting of SEQ ID
NOS:1 - 651, or its complement, provided that the polymorphic sequence

includes a nucleotide other than the nucleotide recited in for said polymorphic sequence, or the complement includes a nucleotide other than the complement of the nucleotide recited in Table 1, column 5; and

- c) determining whether said nucleic acid and said oligonucleotide hybridize;
 whereby hybridization of said oligonucleotide to said nucleic acid sequence indicates
- 24. A method of determining the relatedness of a first and second nucleic acid, the method comprising:

the presence of the polymorphism in said subject.

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- a) providing a first nucleic acid and a second nucleic acid;
- b) contacting said first nucleic acid and said second nucleic acid with an oligonucleotide that hybridizes to a polymorphic sequence selected from the group consisting of SEQ ID NOS:1 651, or its complement, provided that the polymorphic sequence includes a nucleotide other than the nucleotide recited in Table 1, column 5 for said polymorphic sequence, or the complement includes a nucleotide other than the complement of the nucleotide recited in Table 1, column 5;
- c) determining whether said first nucleic acid and said second nucleic acid hybridize to said oligonucleotide; and
- d) comparing hybridization of said first and second nucleic acids to said oligonucleotide,

wherein hybridization of the first and second nucleic acids to said oligonucleotide indicates the first and second nucleic acids are related.

25. The method of claim 24, wherein said oligonucleotide does not hybridize to said polymorphic sequence when said polymorphic sequence includes the nucleotide recited in Table 1, column 5 for said polymorphic sequence, or when the complement

of the polymorphic sequence includes the complement of the nucleotide recited in Table 1, column 5 for said polymorphic sequence.

26. The method of claim 24, wherein the oligonucleotide is between about 10 and about 51 bases in length.

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- 27. The method of claim 24, wherein the oligonucleotide is between about 10 and about 40 bases in length.
- The method of claim 24, wherein the oligonucleotide is between about 15 and about 30 bases in length.
 - 29. An isolated polypeptide comprising a polymorphic site at one or more amino acid residues, wherein the protein is encoded by a polynucleotide selected from the group consisting of: polymorphic sequences SEQ ID NOS:1 651, or their complement, provided that the polymorphic sequence includes a nucleotide other than the nucleotide recited in Table 1, column 5 for said polymorphic sequence, or the complement includes a nucleotide other than the complement of the nucleotide recited in Table 1, column 5.
- 20 30. The polypeptide of claim 29, wherein said polypeptide is translated in the same open reading frame as is a wild type protein whose amino acid sequence is identical to the amino acid sequence of the polymorphic protein except at the site of the polymorphism.
- The polypeptide of claim 29, wherein the polypeptide encoded by said polymorphic sequence, or its complement, includes the nucleotide listed in Table 2, column 6 or

Table 3, column 5 for said polymorphic sequence, or the complement includes the complement of the nucleotide listed in Table 1, column 6.

- 32. An antibody that binds specifically to a polypeptide encoded by a polynucleotide comprising a nucleotide sequence encoded by a polynucleotide selected from the group consisting of polymorphic sequences SEQ ID NOS:1 651, or its complement, provided that the polymorphic sequence includes a nucleotide other than the nucleotide recited in Table 1, column 5 for said polymorphic sequence, or the complement includes a nucleotide other than the complement of the nucleotide recited in Table 1, column 5.
 - 33. The antibody of claim 32, wherein said antibody binds specifically to a polypeptide encoded by a polymorphic sequence which includes the nucleotide listed in Table 1, column 6 for said polymorphic sequence.

34. The antibody of claim 32, wherein said antibody does not bind specifically to a polypeptide encoded by a polymorphic sequence which includes the nucleotide listed in Table 1, column 5 for said polymorphic sequence.

- 20 35. A method of detecting the presence of a polypeptide having one or more amino acid residue polymorphisms in a subject, the method comprising
 - a) providing a protein sample from said subject;

- b) contacting said sample with the antibody of claim 34 under conditions that allow for the formation of antibody-antigen complexes; and
- c) detecting said antibody-antigen complexes,
 whereby the presence of said complexes indicates the presence of said polypeptide.

36. A method of treating a subject suffering from, at risk for, or suspected of, suffering from a pathology ascribed to the presence of a sequence polymorphism in a subject, the method comprising:

- 5
- a) providing a subject suffering from a pathology associated with aberrant expression of a first nucleic acid comprising a polymorphic sequence selected from the group consisting of SEQ ID NOS:1 - 651, or its complement; and

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b) administering to the subject an effective therapeutic dose of a second nucleic acid comprising the polymorphic sequence, provided that the second nucleic acid comprises the nucleotide present in a wild type allele of the sequence polymorphism,

thereby treating said subject.

- 15 37. The method of claim 36, wherein the second nucleic acid sequence comprises a polymorphic sequence which includes the nucleotide listed in Table 1, column 5 for said polymorphic sequence.
- 38. A method of treating a subject suffering from, at risk for, or suspected of suffering from a pathology ascribed to the presence of a sequence polymorphism in a subject, the method comprising:
 - a) providing a subject suffering from a pathology associated with aberrant expression of a polymorphic sequence selected from the group consisting of polymorphic sequences SEQ ID NOS:1 - 651, or its complement; and

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b) administering to the subject an effective therapeutic dose of a polypeptide,

wherein said polypeptide is encoded by a polynucleotide comprising a polymorphic sequence selected from the group consisting of SEQ ID NOS:1 - 651, or by a polynucleotide comprising a nucleotide sequence that is complementary to any one of polymorphic sequences SEQ ID NOS:1 - 651, provided that said polymorphic sequence includes the nucleotide listed in Table 1, column 6 for said polymorphic sequence, thereby treating said subject.

39. A method of treating a subject suffering from, at risk for, or suspected of suffering from, a pathology ascribed to the presence of a sequence polymorphism in a subject, the method comprising:

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- a) providing a subject suffering from, at risk for, or suspected of suffering from, a pathology associated with aberrant expression of a first nucleic acid comprising a polymorphic sequence selected from the group consisting of SEQ ID NOS:1 - 651, or its complement; and
- b) administering to the subject an effective dose of the antibody of claim 34, thereby treating said subject.
- 40. A method of treating a subject suffering from, at risk for, or suspected of suffering from, a pathology ascribed to the presence of a sequence polymorphism in a subject, the method comprising:
 - a) providing a subject suffering from, at risk for, or suspected of suffering from, a pathology associated with aberrant expression of a nucleic acid comprising a polymorphic sequence selected from the group consisting of SEQ ID NOS:1 - 651, or its complement; and
 - b) administering to the subject an effective dose of an oligonucleotide comprising a polymorphic sequence selected from the group consisting of SEQ ID NOS:1 - 651, or by a polynucleotide comprising a nucleotide sequence that is complementary to any one of polymorphic sequences SEQ

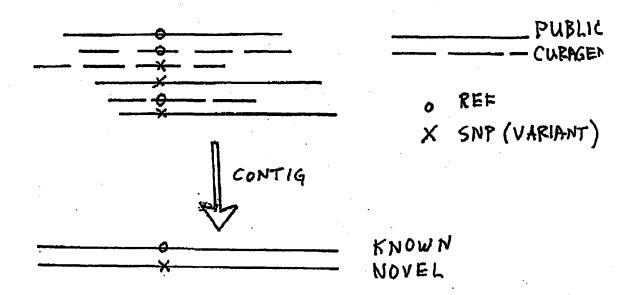
ID NOS:1 - 651, provided that said polymorphic sequence includes the nucleotide listed in Table 1, column 6 for said polymorphic sequence,

thereby treating said subject.

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- An oligonucleotide array, comprising one or more oligonucleotides hybridizing to a first polynucleotide at a polymorphic site encompassed therein, wherein the first polynucleotide is chosen from the group consisting of:
 - a) a nucleotide sequence comprising one or more polymorphic sequences SEQ
 ID NOS:1 651;
 - b) a nucleotide sequence that is a fragment of any of said nucleotide sequence,
 provided that the fragment includes a polymorphic site in said polymorphic sequence;
 - c) a complementary nucleotide sequence comprising a sequence complementary to one or more polymorphic sequences SEQ ID NOS:1 -651; and
 - d) a nucleotide sequence that is a fragment of said complementary sequence, provided that the fragment includes a polymorphic site in said polymorphic sequence.
- 20 42. The array of claim 41, wherein said array comprises 10 oligonucleotides.
 - 43. The array of claim 41, wherein said array comprises at least 100 oligonucleotides.
 - 44. The array of claim 41, wherein said array comprises at least 1000 oligonucleotides.

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F14. 1

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<210> 737
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Ile Leu Pro Ser Gly Leu Ala Phe Ile Ser Thr Gly Leu Lys
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Phe Tyr Leu Ala Met Asn Glu Glu Gly Lys Leu Tyr Ala Lys
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<210> 739
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<223> cSNP translation
Gly Leu Asp Gln Lys Arg Ile Lys Tyr Val Val Gly Glu Leu
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Ala Gly Leu Gly Gln Val Arg Leu Ile Val Gly Ile Leu Leu
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Thr Leu Thr Ser Val Gly His Gln Ser Val Thr Pro Gly Glu
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 Gly Gln Lys Thr Leu Thr Pro Val Gly Tyr Gln Ser Val Thr
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 Thr Glu Ile Thr Gly Ala Thr Thr Met Thr Ser Val Gly His
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 Gly Val Tyr Ile Leu Thr Tyr Asn Thr Ser Gln Tyr Asp Thr
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Pro Asp Pro Arg Glu Ala Cys Gly Ser Ser Ser Tyr Val
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Ala Lys Asp Met Asn Gly Thr Ser Leu His Gly Lys Ala Ile
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Ile Gln Tyr Thr Tyr Leu Gly Gly His Val Cys Leu Ser Ala
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Leu Asp Pro Asn Asn Pro Asp Ala Asn Trp Ile His Ala Arg
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Cys Leu Thr Glu Arg Gln Ser Lys Ile Trp Phe Gln Asn Arg
1
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Pro Tyr Arg Ile Ala His Ala Val Ile Lys Ala His Ala Arg
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<223> cSNP translation
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Lys Arg Lys Lys Glu Val His Ala Thr Ser Pro Ala Pro Ser
                5
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<210> 756
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Ile Lys Asn Glu Ala Arg Leu Pro Cys Leu Pro Thr Pro Gly
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Ala Arg Lys Phe Phe Glu Asn Leu Pro Asp Gly Thr Trp Asn
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Phe Ser Tyr Ser Ala Ser Ser Thr
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Trp Glu Arg Phe Val His Arg Glu Asn Gln His Leu Val Ser
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Ala Pro Pro Gly Ala Tyr His Gly Ala Pro Gly Ala Tyr Pro
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Val Met Pro His Ser Ser Glu His Lys Thr Ala Gln Pro Asn
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Thr Cys Val Val Glu His Thr Gly Ala Pro Glu Pro Ile Leu
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Val Gly Phe Leu Val Gly Ile Val Leu Ile Ile Met Gly Thr
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Gln Ser Val Val Ser Cys Ala
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<223> cSNP translation
Pro Gly Pro Thr Val Arg Ala Gly Glu Asn Val Thr Leu Ser
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Cys Ser Gly Val Trp Gly Ala Asp Thr Glu Glu Arg Leu Val
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Leu Leu Arg His Glu Trp Gln Gly Tyr Gln Lys Trp Val Arg
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 Gly Phe His Tyr Gly Val Leu Ala Cys Glu Gly Cys Lys Gly
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 Asp Gly Pro Glu Gln Ala His Arg Gln Arg Arg Gln Arg
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<223> cSNP translation
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Ile Leu Asp Thr Ala Gly His Glu Glu Tyr Ser Ala Met Arg
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Phe Gly His Gln Glu Asn Ser Gln Asn Glu Glu Ile Leu Asn
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Gln Pro Gln Ala Arg Gln Glu Glu Gln Val Arg Val Val Arg
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<211> 14
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Asn Ala Val Lys Ala Glu Thr Arg Gln Gln Phe Arg Ser Leu
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Asp Asp Asp Ala Pro Arg Pro Ser Gln Phe Glu Glu Asp
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His Val Leu Arg Met His Gly Tyr Arg Ala Pro Gly Glu Gln
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<223> cSNP translation

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Glu Thr Gln Leu Gly Thr Leu Ala Gln Phe Pro Asn Thr Leu
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<210> 788
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Glu Ala Met Glu Arg Phe Gly Glu Asp Glu Gly Phe Ile Lys
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Glu Glu Glu Lys Pro Leu Ala Arg Asn Glu Phe Gln Arg Gln
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Asp Ala His Phe Asp Glu His Glu Arg Trp Thr Asn Asn Phe
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Arg Trp Thr Asn Asn Phe Thr Glu Tyr Asn Leu His Arg Val
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Asp Ser Gly Leu Leu Gln Cys Lys Leu Ala Leu Leu Ser Val
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Ala Phe Val Val Leu Ala Leu Gln Phe Leu Ser His Asp Pro
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Thr Gly Lys Ile Gln Arg Thr Lys Leu Arg Asp Lys Glu Trp
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Leu Arg Ile Gln Cys Leu Cys Arg Lys Gln Ser Ser Lys His
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Leu Glu Lys Ile Gln Pro Met Thr Gln Asn Gly Gln His Pro
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Trp Met Ile Phe Val Val Ile Ala Ser Val Phe Thr Asn Gly
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Ser Ile Asn Leu Phe Ser Gly Ile Phe Phe Leu Thr Cys Met
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<223> cSNP translation
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Glu Ser Asn Thr Thr Gly Thr Thr Ala Phe Ser Met Pro Ser
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Val His Pro Val Arg Pro Leu Arg Leu Glu Ser Phe Ser Ala
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Arg Gly Ala Arg Pro Gly Pro Arg Val Pro Lys Thr Leu Val
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 Glu Leu Tyr Arg Asp Ile Leu Gln His Leu Arg Asp Glu Ser
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 <223> cSNP translation
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 Tyr Val Phe Thr Asp Gln Leu Ala Ala Val Pro Arg Val Thr
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<223> cSNP translation
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Leu Ser Val Leu Glu Val Gly Ala Tyr Lys Arg Trp Gln Asp
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<210> 811
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<223> cSNP translation
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Val Asp Val Asp Met Glu Ile Arg Asp His Val Gly Val Glu
<210> 812
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<223> cSNP translation .
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Phe Gly Thr Leu His Pro Ser Phe Tyr Gly Ser Ser Arg Glu
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<223> cSNP translation
Glu Gly Asp Phe Tyr Tyr Met Gly Gly Phe Phe Gly Gly Ser
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Gly Gly Ser Val Cln Glu Met Gln Arg Leu Thr Arg Ala Cys
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WO 01/48245 PCT/US00/35346 <211> 14

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 <223> cSNP translation
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 <223> cSNP translation
· <400> 816
Lys Asp Phe His Lys Asp Met Leu Lys Pro Ser Pro Gly Lys
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 <210> 817
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 Thr Asn Asn Cys Tyr Arg His Ala Ile Val Thr Thr Ser Ile
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 Gly Phe Val Val Phe Ser Ser Leu Gly Tyr Met Ala Gln Lys
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 <210> 819
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 <212> PRT
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 <222> (7)...(0)
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Met Asp Glu Ala Ala Arg Pro Glu Ala Trp Asp Ser Tyr Arg
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<210> 820
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<223> cSNP translation
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Ser Pro Gln Ser Ser Ala Arg Gly Lys Pro Ala Met Ser Tyr
<210> 821
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<212> PRT
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<222> (7)...(0)
<223> cSNP translation
<400> 821
Leu Glu Asp Leu Ala Gly Trp Lys Glu Leu Phe Gln Thr Pro
<210> 822
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Val Gln Asp Ile Leu Arg Leu Glu Met Pro Ala Ser Lys Ile
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                 5
                                    10
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Lys Val Leu Asp His Trp Cys Ile Met Thr Ser Glu Glu Glu
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Gln Glu Val His Gly Pro Tyr Pro Asp Ser Ser Phe Leu Thr
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<210> 826
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<222> (7)...(0)
<223> cSNP translation
Gln Glu Val His Gly Pro Ile Pro Asp Ser Ser Phe Leu Thr
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<210> 827
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<223> cSNP translation
Glu Leu Cys His Glu Lys Gly Ile Leu Glu Lys Tyr Gly His
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<210> 828
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<212> PRT
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<223> cSNP translation
<400> 828
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Asn Asn Asn Leu Arg His Thr Asp Glu Met Phe Trp Asn His
<210> 829
<211> 14
<212> PRT
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<221> VARIANT
<222> (7)...(0)
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Gly Met Ala Ser Ser Cys Ser Val Gln Val Lys Leu Glu Leu
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<210> 830
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Pro Ser Ile Phe Ile Tyr Arg His Thr Ala Ser Gly Lys Thr
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<210> 831
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Pro Pro Pro Pro Gly Ala Pro Gly Gly Ser Gln Asp Thr
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<223> cSNP translation
<400> 832
Arg Ala Ala Leu Glu Arg Gly Lys Ala Ile Glu Lys Asn Leu
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<220>
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<223> cSNP translation
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Thr Gly Gly Leu Leu Leu Arg Leu Ala Leu Met Leu Gln Leu
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<223> cSNP translation
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Asp Ser Ser Ser Asn Gly Lys Ala Lys Asn Pro Pro Gly
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<223> cSNP translation
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Leu Glu Pro Gln Trp Tyr Ser Val Leu Glu Lys Asp Ser Val
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<210> 836
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<223> cSNP translation
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Gln Lys His Ser Ser Gly Xaa Ser Asn Thr Ser Thr Ala Asn
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Trp Gly Thr Glu Asp Asp Ala Thr Gln Ser Tyr His Asn Gly

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Phe Ser Lys Leu Leu Gly Pro Leu Ser Ala Lys Lys Tyr Leu
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 Thr Ser Lys Ile Leu Phe Phe Ser Gln Gly Ser Glu Ile Ala
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 Tyr Val Gln Pro Pro Glu Val Ile Gly Pro Met Arg Pro Glu
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<222> (7)...(0)
<223> cSNP translation
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Gly Pro Asp Gly Gln Glu Val Asp Pro Pro Asn Pro Glu Glu
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Lys Gly Glu Gln Lys Arg His Glu Lys Ile Ser Ala Ser
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Ala Pro Gln Glu Glu Glu Ala Ser Lys Glu Lys Glu Glu
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Lys Val Val Gln Ser Pro Gln Ser Leu Val Val His Glu Gly
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<210> 846
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Leu Asn Cys Ser Tyr Glu Met Thr Asn Phe Arg Ser Leu Leu

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<210> 847

<400> 846

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<223> cSNP translation
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Leu Asp Lys Lys Glu Leu Ser Ser Ile Leu Asn Ile Thr Ala
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<210> 849
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<223> cSNP translation
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Glu Tyr Val Val Gly Ala Pro His Leu Glu Leu Asp Pro Gly
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                                    10
<210> 850
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<223> cSNP translation
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Phe Phe Lys Arg Asn Arg His Thr Pro Gly Arg Arg
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<223> cSNP translation
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Thr Ile Gln Pro Pro Arg Glu
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<210> 852
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Gly Ile Val Gly Gln Lys Gly Arg Pro Trp Leu Pro Arg Thr
                5
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<223> cSNP translation
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Gly Gly Lys Met Gly Gly Arg Lys Arg Leu Gln Lys
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Tyr Ser Ser Tyr Gly Gln Ser Leu Phe Thr Val Leu Trp Trp
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<222> (8)...(0)
<223> cSNP translation
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Glu Gln Leu Arg Arg Gln Leu Asp Pro Leu Arg Thr Ala His
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<210> 856
<211> 14
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<220>

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<223> cSNP translation

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 <223> cSNP translation
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 Arg Tyr Leu Asp Trp Ile Leu Trp Ala His Gln Arg
 <210> 862
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 <212> .PRT
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 <222> (8)...(0)
 <223> cSNP translation
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 Ala Glu Leu Arg Leu Leu Arg Ala Gln Val Lys Ser Gly Ala
                 5
                                     10
 <210> 863
 <211> 14
 <212> PRT
 <213> Homo sapiens
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 <223> cSNP translation
 Ala Glu Leu Arg Leu Leu Arg Ala Gln Val Lys Ser Gly Ala
                  5
 <210> 864
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 <212> PRT
 <213> Homo sapiens ·
 <220>
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 <222> (8)...(0)
 <223> cSNP translation
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 Pro His Cys Arg Pro Gly Ala Trp Pro Ala Thr Glu Arg Gly
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 <210> 865
 <211> 14
 <212> PRT
 <213> Homo sapiens
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<221> VARIANT
<222> (8)...(0)
<223> cSNP translation
<400> 865
Ile His Phe Glu Asp Tyr Gly Val Leu Gly His His Gln Leu
    5
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<210> 866
<211> 14
<212> PRT
<213> Homo sapiens
<220>
<221> VARIANT
<222> (7)...(0)
<223> cSNP translation
<400> 866
Asn Phe Ile Leu Ala Cys Pro Arg
<210> 867
<211> 14
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<223> cSNP translation
<400> 867
Arg Glu Lys Leu Arg Asn Phe His Phe Ser Met Ser Arg
<210> 868
<211> 14
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<223> cSNP translation
Leu Leu Leu Leu Leu Arg Arg Pro Ala Gln Pro Gln Leu
           . 5
<210> 869
<211> 14
<212> PRT
<213> Homo sapiens
<220>
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<222> (7)...(0)
<223> cSNP translation
<400> 869
Lys Arg Val Ala Gly Gly Leu Arg
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  <400> 870
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  <210> 871
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  <400> 871
  Ser Ser Gly Arg Pro Thr Gly Tyr Cys Leu Gln Leu Gln Gln
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  <223> cSNP translation
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  Gln Arg Ser Ile Ser Ala Asp
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  <212> PRT
  <213> Homo sapiens
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  <222> (8) ...(0)
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 Arg Ala Pro Val Ile Leu Gly Pro Pro Thr Thr Cys Ser Ser
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<222> (7)...(0)
<223> cSNP translation
<400> 874
Met Gly Leu Ser Gly Phe Leu Thr Gly Pro Pro Pro Pro Gly
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<210> 875
<211> 14
<212> PRT
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<221> VARIANT
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<223> cSNP translation
<400> 875
Leu Met Gly Leu Ser Gly Phe Leu Thr Gly Pro Pro Pro
                                   1.0
<210> 876
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<212> PRT
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<222> (7)...(0)
<223> cSNP translation
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Pro Arg Thr Pro Ala Glu Pro Pro Pro Leu Gly Arg Gln Ala
                5
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<210> 877
<211> 14
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<221> VARIANT
<222> (7)...(0)
<223> cSNP translation
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Gly Thr Gly Asp Trp Arg Glu Pro Gly Ala Ala Ser Glu Arg
<210> 878
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<212> PRT
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<222> (9)...(0)
<223> cSNP translation
Gln Gly Arg Gln Ser Lys Gly Leu Arg Arg Arg Thr Trp Pro
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<210> 879

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<212> PRT
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<222> (8)...(0)
<223> cSNP translation
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Lys Cys Lys Cys Ser Arg Lys Asp Pro Arg Ser Ala Thr Ala
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<211> 14
<212> PRT
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<221> VARIANT
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<223> cSNP translation
<400> 880
Cys Lys Cys Ser Arg Lys Asp Pro Arg Ser Ala Thr Ala Thr
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